Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

The National Healthy Childhood Programme

Standard Operating Procedure for Maternity Hospitals/Units & Primary Care Services Delivering the National Newborn Bloodspot Screening Programme (NNBSP)

Is this document a:

Policy	**Procedure**	Protocol	Guideline
	x		

HSE National Acute Hospital Division *– Maternity Hospitals/Units*
HSE Primary Care Division *– Public Health Nursing, Community Midwives, Self Employed Community Midwives*
National Newborn Bloodspot Screening Programme Governance Group

Title of PPPG Development Group:	National Newborn Bloodspot Screening Programme Governance Group
Approved by:	National Head of Operations Health And Wellbeing National Head of Operations Primary Care Community Healthcare Organisations Child Health Leads Primary Care Management Teams for each CHO/Local Health Office Management Teams for Maternity Hospital /Units, Hospital Groups (This approval obtained in August 2016 but delayed implementation of SOP. Outline of PPPG steps have not changed)
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PART A: Outline of PPPG Steps

Outline the step by step process to follow, algorithm, process flow chart, or (SOP) which has been developed using the HSE National Framework for developing PPPGs. *(Part B should be completed first to develop the PPPG and then the core PPPG steps that have been developed are inserted in Part A).*

Standard Operating Procedure for Maternity Hospitals/Units & Primary Care Services Delivering the National Newborn Bloodspot Screening Programme

2.7 Outline of PPPG steps/recommendations:

Procedure for taking initial newborn bloodspot screening sample

Stage One: Notification of birth to CHOs and requests for newborn bloodspot screening sample to Public Health Nursing services/LHOs

2.7.1 Notification of birth to CHO

2.7.1.1 Maternity Hospitals/Units, both within and outside of the Republic of Ireland (ROI), notify the CHO of all babies born and who reside in the designated CHO (Full Birth Notification – BNF-01). A copy of the BNF-01 is sent to:

- Registrar of births – demographic information
- Director of Public Health – clinical and demographic information
- National Perinatal Reporting System (NPRS) – clinical information
- Hospital/Unit copy – clinical and demographic information

It is noted that the copy for the Director of Public Health, in practice today, now goes to the CHO, community child health services/office or Director of Public Health Nursing Office.

2.7.1.2 The Maternity Hospital/Unit sends the Full Birth Notification to the local Child Health Office either electronically or in paper format[1]:

- For a number of CHOs (Dublin Areas), the Child Health Information Systems (CHIS) office receives the Full Birth Notification and inputs the information into the Child Immunisation/Parent Held Record (PHR) System. This information is electronically transferred on a daily basis to the DPHN Office in the LHO (12 midnight). The DPHN Office/LHO set up a Child Health Record and forwards this to the Primary Care Team (PCT) RPHN.
- For other CHOs, the Full Birth Notification information is received at one or many locations, the DPHN Office, the Birth Notification Office, the Child Health Office and/or Primary Care Unit Management (PCUM)[2]. By agreement, the DPHN Office and Birth Notification/PCUM, input the information into the Child Immunisation/Child Health/PHR System. The RPHNs set up a Child Health Record within local guidelines which includes newborn screening.

[1] Birth Notifications should be made electronically as far as possible e.g. CHIS or secure email or Shared Drive systems and/or paper format. The practise of using fax should be minimised or discontinued.

[2] It is recommended that all child health databases and birth notifications be coordinated and managed by a Child Health Office for the CHO/LHO.

- The development and roll out of the Maternal and Newborn Clinical Management system (MN-CMS) and National Immunisation and Child Health Immunisation System (NICHIS) will standardise the collection of data further.

2.7.2 Maternity Hospital/Unit Births and NNBSP

2.7.2.1 The Maternity Hospital/Unit is responsible for completing the newborn bloodspot screening test (NBST) for all babies on the wards between 72 and 120 hours following the birth and delegating duties to the relevant Midwives/Nurses. Where babies are being discharged before 72 hours, the Maternity Hospital/Unit is responsible for notifying the relevant CHO/designated LHO Public Health Nursing Service of the requirement to carry out the NBST.

2.7.2.2 The Maternity Hospital/Unit:

- Maintains a birth register for all babies born in the hospital/unit.
- Issues the **Unique Perinatal Identifier (UPI)** number for all babies born in the Hospital/Unit (Appendix VI). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- Maintains a NBS Register for babies who have NBS completed while in Hospital.
- Delegates duties for maintaining the NBS Register to a designated officer.

2.7.2.3 The Maternity Hospital/Unit forwards the NBSC taken within the requisite timeframe (72-120 hours) to the NNBSL and confirms to the DPHN the date and time NBS sample was obtained from the baby.

2.7.2.4 If the sample is not taken within the Hospital before discharge, the DOMN/designated staff member is responsible for ensuring that:

- The designated officer in the CHO/LHO is notified of the birth.
- The DPHN[3] /Child Health Office[4] is informed that the baby has been discharged prior to the sample being carried out and is notified of the request for NBS[5]. The Hospital/Unit should provide two contact details of Parent(s)/legal Guardian(s) including mobile telephone number if known. Where a Parent(s)/legal Guardian(s) and baby are staying at a temporary address this should be made clear on the NBS notification.

2.7.2.5 The Maternity Ward within the Maternity/Unit Hospital has responsibility for:

- Completing an **Early Discharge Summary/Edited Birth Notification** for all babies that require the NBS sample to be taken in the CHO/LHO and forwarding it to the designated DPHN.
- The date the newborn bloodspot screening sample (NBSS) is required to be taken should be clearly written on the Discharge Summary/Edited Birth Notification.

[3] For the purposes of this document the point of contact between Hospital and Primary Care for NBS is the office of the DPHN. Some CHOs have a Liaison PHN acting on behalf of the DPHN, coordinating birth notifications and NBS requests, screening discharges from hospitals and maintaining birth and NBS registers for the CHO/LHO for audit and collation of key performance indicators.

[4] Some CHOs/LHOs the child health office/immunisation/central referrals office coordinates birth notifications and NBS requests on behalf of the office of the DPHN.

[5] NBS notifications should be made electronically as far as possible e.g. secure email or shared drive systems and/or paper format. The practice of using fax should be minimised or discontinued.

- Completing the following information on the NBSC: The **Baby's UPI number; Time of Birth; Date of first feed**; **Type of Feed.** The mother will be given the *HSE Parent/Guardian Information Leaflet* (Appendix VII).
- Notifying the DPHN if the child or mother is **infected with a pathogen** e.g. HIV or Hepatitis B**,** which would result in the card not being exempt from the **Biohazard packaging regulations**.
- Giving the mother the **Discharge Checklist: Baby.** The mother will give this discharge summary to the PCT RPHN (Appendix VIII).

2.7.2.6 In the case of early discharges or where the Maternity Hospital/Unit wish to observe the baby, the DOMN/designated staff member may issue an appointment for the baby to return to the hospital for follow up during the designated timeframe for which the NBS can be undertaken.

2.7.2.7 The Maternity Hospital/Unit may request that mother and baby return to the Hospital/Unit at weekends/bank holidays/extended holiday periods, to facilitate early discharge and where the Public Health Nursing Service does not provide a weekend service.

2.7.3 Hospital /Unit Transfer to Tertiary Hospital

2.7.3.1 It may be necessary for a baby to be transferred to a tertiary hospital for continuing medical and/or surgical treatment before the NBSS has been taken.

2.7.3.2 The nurse/midwife responsible for the transfer of a baby from a maternity unit/hospital to a tertiary referral paediatric unit must inform the receiving unit that the NBST is required and give them the baby's UPI.

2.7.3.3 The receiving paediatric unit must have written procedures for:

- Performing the test between 72 and 120 hours after birth.
- Sending the newborn bloodspot screening sample (NBSS) to the NNBSL.
- Recording the results in the baby's medical records.
- Informing the Maternity Hospital/Unit of the NBST results.

2.7.4 Hospital/Unit Notification of NBS Request to PHN Service

2.7.4.1 Maternity Hospital/Unit, within and outside the ROI, notify the request to carry out NBS to the Public Health Nursing Service for the designated LHO. This may be part of the pending discharge (Discharge Summary/Edited Birth Notification and/or Telephone contact/Fax)[6].

- In the case of a family and baby relocating and/or returning to reside in ROI (from outside ROI) prior to the NBST, it is the responsibility of the Maternity Hospital/Unit to inform the DPHN for the designated LHO that the baby and family is relocating/returning within the ROI, within the timeframe for the sample to be taken (72-120 hours).
- It is the responsibility of the Maternity Hospital/Unit to inform the relevant Public Health Nursing Service/LHO in the case of a family and baby discharging to a temporary address (e.g. grandmother) prior to returning to the address of their main residence.
- Maternity Hospitals/Units can use the Health Atlas address finder facility to help check the correct LHO to write on the NBSC. https://finder.healthatlasireland.ie/
- Maternity Hospitals/Units are reminded that it is important for the screening process that both temporary and permanent addresses are flagged on the NBSC (on separate

[6] NBS Notifications should be made electronically as far as possible e.g. Secure email or Shared Drive systems, and/or paper format. The practice of using fax should be minimised or discontinued.

sheet if appropriate) and the 36 hour birth notification to the LHO is completed, in order to ensure that immediate follow up is facilitated and to clarify where baby will reside on a permanent basis and avail of other child health services.

2.7.4.2 Where a request for NBS comes from a Maternity Hospital/Unit to an incorrect LHO, the DPHN/LHO must return the NBS request to the Maternity Hospital/Unit. The Maternity Hospital/Unit must correct this information and send the request to the correct LHO and notify the NNBSL of the change of LHO.

2.7.4.3 Northern Ireland (NI) Maternity Hospitals/Units will contact the Public Health Nursing Service/LHO and request:

- The Public Health Nursing Service Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.4.4 Please note that in the case of babies born outside the jurisdiction, the Parent(s)/legal Guardian(s) are equally responsible for ensuring that the baby is screened. If the Parent(s)/legal Guardian(s) notify the Public Health Nursing Service of the birth on their return to this jurisdiction, the Public Health Nursing Service is responsible for ensuring that the sample is carried out in this jurisdiction. The RPHN/RM may seek the advice of the NNBSL and/or Family GP.

2.7.5 Self Employed Community Midwife (SECM) notification of birth and NBS status to PHN service

2.7.5.1 The SECM is responsible for undertaking NBS for all babies in their care who require the NBST. The NBS sample will be taken between 72 hours and 120 hours following the birth.

2.7.5.2 The SECM will contact the DPHN/LHO and request:

- The Public Health Nursing Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.5.3 The SECM will notify the DPHN for the designated LHO of the home birth and pending discharge from SCEM service (Discharge Summary/Edited Birth Notification).

2.7.6 Midwifery Groups notification of NBS status to PHN service

2.7.6.1 Midwifery Groups contracted by the Parent(s)/legal Guardian(s) to provide postnatal care are responsible for undertaking NBS for all babies in their care who require the NBST. The NBS sample will be taken between 72 and 120 hours following the birth.

2.7.6.2 Midwifery Groups will contact the DPHN for the designated LHO and request:

- The DPHN to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.

- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.6.3 The Midwifery Group will notify the DPHN for the designated LHO that the NBS sample has been taken and the pending discharge from the Midwifery groups service.

2.7.7 General Practitioner notification of the NBS status to PHN service

2.7.7.1 In the event that a GP undertakes the NBST, they must ensure the NBS sample is taken between 72 hours and 120 hours following the birth.

2.7.7.2 The GP will contact the DPHN/LHO and request:

- The Public Health Nursing Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL

2.7.7.3 The GP will notify the DPHN for the designated LHO that the NBST has been taken.

2.7.7.4 In the case of Traveller babies, the baby may have moved out of the area before the NBST has been carried out. The DPHN must be informed of the circumstances as early as possible so that alternative arrangements may be made.

2.7.8 Self-referral to PHN service and NBS status

2.7.8.1 Where a Parent(s)/legal Guardian(s) presents to the Public Health Nursing Service and notification of the baby's birth and requirement for newborn bloodspot screening has not been communicated from hospital services, the Public Health Nursing Service will provide the required service. This situation may arise for babies born outside of the ROI.

Stage Two: DPHN Office and Notification to designated PCT PHN

2.7.9 Notification to the PCT PHN

2.7.9.1 The DPHN/Designated Officer will

- Ensure on receipt of request for NBS via **Early Discharge Summary/Edited Birth Notification** and/or telephone contact, that the relevant details of the request are correct, including the UPI number for the baby (Appendix VI).
- Ensure that the full details of the baby are correct, including details of the mother and baby who have an infection such as HIV or Hepatitis B that requires specific consideration (See 2.7.15.5 and2.7.17.7) are highlighted to the practitioner.
- When a NBS request is for an incorrect LHO, the DPHN/Designated Officer will notify the Maternity Hospital/Unit of the error immediately.
- Issue a UPI number to Hospital/Unit and SECM, Midwifery Group on request for babies born outside of ROI and home births (Appendix IX).
- Have access to a full electronic Birth Register. The Immunisation/PHR/Child Health Information System is the electronic birth register for the LHO. The Child Health Office coordinates all immunisation and child health databases.
- In the short term, for some LHOs, "fields" from the Immunisation System/PHR/Child Health Information System are extracted and used to populate the NBS Register. For

some LHOs, the Immunisation System/PHR/Child Health Information System is the birth register and the NBS register as it has the facility to enter the required data.

2.7.9.2 The DPHN or Designated Officer will:

- Receive the data extract from the Immunisation/PHR/Child Health Information System.
- Input the additional information on the NBS Register. (Appendix X)
- Notify the designated Primary Care Team (PCT) Registered Public Health Nurse (RPHN)/Registered Midwife (RM)/Registered Nurse (RGN) of the request for the NBS by email or fax.

2.7.9.3 The email will generate an automatic acknowledgement of receipt of notification. Where email is not used the RPHN/RM/RGN acknowledges receipt of the request for Newborn Bloodspot Screening via telephone or fax[7].

2.7.9.4 During the normal working week, the RPHN/RM/RGN contacts the Parent(s)/legal Guardian(s) and arranges an agreed time and place for NBS to be carried out. This may be a:

- Home visit.
- Centre based visit if this is more convenient for the Parent(s)/legal Guardian(s) and/or RPHN/RM/RGN.

2.7.9.5 At weekends and bank holiday weekends:

- For some LHOs, where there is a weekend/bank holiday period service for newborn babies, the Designated Officer informs the weekend Nurse by 12:00 midday on Friday that a NBSS is requested for the baby.
- For some LHOs, where there is no Public Health Nursing Service weekend/bank holiday period service, the Maternity Hospital /Unit requests that the mother, with her baby, return to the Hospital/Unit. The NBST can be performed either in the postnatal ward or at a planned NBS clinic. The Public Health Nursing Service will be informed by the Hospital/Unit of the birth and that the NBSS was taken, on the following Monday/Tuesday after weekend/bank holiday period.
- For some LHOs, where the Maternity Hospital/Unit provides early discharge/domino schemes, the Hospital/Unit requests that the mother, with her baby, return to the Hospital/Unit and/or Community Midwife for the NBS to be undertaken. The NBSS can be performed either in the postnatal ward or at a planned NBS clinic. The Public Health Nursing Service will be informed of the birth and that the NBSS was taken on the following Monday/Tuesday after weekend/bank holiday period.
- As per local arrangements, additional special arrangements can be organised for extended holiday periods/ urgent requests from NNBSL, to ensure the NBSS is delivered from LHO to the NNBSL (For example a courier arrangement).

2.7.9.6 Where a family and baby are resident temporarily in the LHO immediately following birth (e.g. with Grandmother) evidence of newborn bloodspot screening must be obtained. This may take the form of email or written results or a written record in the Child Health Record (CHR). Screening must be carried out if the baby has not been screened. Please note the baby will need to be added to the relevant Child Health Database (Immunisation/PHR/NBS Register).

[7] The practise of using fax should be minimised or discontinued.

2.7.9.7 Where a baby under one year of age has moved into the LHO (temporary or permanent), evidence of NBS must be obtained. This may take the form of email or written results or a written record in the CHR. Where no proof of testing is available it should be assumed that the baby is untested. Screening a child who has not been screened, testing should be discussed with the NNBSL, and where relevant the GP, together with the parent. If recommended for testing, screening must be arranged. In general, all conditions are eligible for testing in unscreened children under one year except Cystic Fibrosis where the screening test is invalid from 6 weeks after birth.

2.7.9.8 Where a baby, up to one year of age, enters the country and has not received NBS for all the conditions screened in the Irish NNBSP in the baby's country of origin, clarification will be sought from the NNBSL and the baby's family GP regarding the required screening for the baby. In general, all of the conditions (except Cystic Fibrosis, where the screening test is invalid from 6 weeks after birth) should be screened for in unscreened children under one year.

2.7.9.9 The decision to screen children greater than one year is a local decision, made by the clinician and family GP and is dependent on the family history and the country of origin of the parents. In such circumstances the NNBSL should be contacted for advice in advance of sample taking.

2.7.9.10 Red Weather alerts: In the event of a red weather alert due to adverse weather conditions, the sample taker in conjunction with the ADOM/ADPHN must assess the risk of travel of either the PHN or the parent and baby against the risk of delayed screening. Further advice can be sought from the laboratory on such occasions.

Stage Three: Procedure for NBS sample taking

2.7.10 Information Giving

2.7.10.1 Parent(s)/legal Guardian(s) should have received information on the NNBSP antenatally in the 3rd trimester and at discharge from hospital/units.

2.7.10.2 All Parent(s)/legal Guardian(s) will be given:

- The NNBSP *HSE Parent/Guardian Information Leaflet* (Appendix VII) if they have not already received this information. Information Leaflets can be obtained on the website: *www.newbornscreening.ie* including information leaflets in a selection of other languages.
- The Parent Information Copy (Appendix XI) of the *Newborn Bloodspot Screening Card* (NBSC) (Appendix XII) which contains information about the NNBSP.

2.7.10.3 All Parent(s)/legal Guardian(s) will receive the following information:

- The conditions which are being screened for and the reason for screening.
- Advise that further samples may be required.
- The NBSC will be retained for 10 years (Note: NBSCs area currently retained indefinitely pending decision by the Minister of Health).

2.7.10.4 The Parent who signs the NBSC must be the legal Guardian. Please note:

- If the Parents are married, both Parents are legal Guardians and either can sign the NBSC.
- If Parents are not married, the mother is automatically the legal Guardian of the baby and can sign the NBSC. The unmarried father may not be automatically the guardian of the baby, and may not be in a position within the timeframe for the NBS to have legal responsibility to sign the NBSC.

2.7.11 Completing the Newborn Screening Card

2.7.11.1 The Sample Taker completes the details required for the NBSC:

- *Baby's Unique Perinatal Identifier (*UPI)
- *Babies demographic details:* Baby's surname, first name, address including postal code/eircode.
- *Place of birth: Hospital or Community.*
- *Babies healthcare record number/transfer to another hospital number*
- *Mother's surname*
- *Local Health Office*
- *Location sample taken:* Please tick the relevant box 'Community' or 'Hospital'.
- If the NBSS is taken in a hospital, other than the birth hospital, please enter the hospital in which the sample is actually taken and the specific Children's/General Hospital/Department/Ward.
- *GP's Name.*
- *Gestational age; time of birth; date of birth, gender.*
- Birth Weight; Rank (For multiple births e.g. Twin 1 Twin 2); date of first feed.
- *Blood Transfusion*; Date and time of first transfusion, date and time of last second transfusion.
- *Type of Feed:* breast, artificial, Total Parenteral Nutrition (TPN), IV fluids, soya/lactose free. Some sample-takers have ticked the incorrect box for **Type of Feed.** The box on the **LEFT** of the **title** of the feed must be ticked.
- *Comments: Family History, Beutler, Meconium Ileus.*
- *Date and time of collection, repeat specimen (yes or no).*
- *Sample taker name and phone contact number:* This requires the phone number of the **sample taker** (or of other individual/centre as determined locally by the DPHN) to enable the sample taker to be contacted directly should this be required. The Parent(s)/legal Guardian(s) phone number is **NOT** to be entered here.
- *Parent(s)/legal Guardian(s) Preferred language:* This field should always be completed – for English, or any other language, as an interpreter could be required to communicate possible positive results and the need to attend a hospital for further samples.
- *Parent(s)/legal Guardian(s) consent signature.*
- *Please note details on the reverse side of the NBSC will be filled in by the laboratory.*

2.7.11.2 The Sample Taker when completing the NBSC ensures that:

- The top information sheet is removed and given to the Parent(s)/legal Guardian(s) together with the parent copy (2nd sheet of NBSC) at the time of sampling.
- Parent(s)/legal Guardian(s) will be informed that results will be issued to the maternity hospitals/units and LHOs only. Results will not be issued to Parent(s)/legal Guardian(s) over the telephone.

- Further information is available on www.newbornscreening.ie or by e-mailing info.newbornscreening@cuh.ie

2.7.11.3 Where there is an error in recording the 'type of feed' identified, the NNBSL needs to be notified immediately by emailing info.newbornscreening@cuh.ie identifying the service location, stating the baby's clinical details and the amendment to be made.

2.7.12 Consent

2.7.12.1 The Parent(s)/legal Guardian(s) by signing the NBSC confirm that:

- He/she received, read and understood the *HSE Parent/Guardian Information Leaflet*.
- The details of the baby on the NBSC are correct.
- They consent to the screening blood sample being taken and a repeat sample if required
- They agree to the storage of the NBSC as per current Department of Health recommendations.

Who can give consent?

- Married parents – if the mother and father are married at the time of birth then either can give consent to screening as they are joint guardians of the infant as per Section 6 of the *Guardianship of Infants Act 1964*
- Unmarried parents – if the mother and father are unmarried at the time of birth only the mother can give consent as per Section 6(4)of the *Guardianship of Infants Act 1964*
- If the mother is unavailable to sign the consent, i.e. through illness or hospital transfer, the unmarried father cannot sign the consent. In these cases, the sample taker should make every effort to contact the mother to get verbal consent and to document this in the relevant clinical notes/child health record. This may include liaising with the mother's medical team to obtain developments on her condition and position to provide consent for the newborn bloodspot screening sample to be taken.
- If the mother is not contactable, for example due to severe inpatient medical illness, then the HSE must act in the best interest of the infant which would be to take the newborn bloodspot screening sample and inform the mother as soon as possible as to the decision taken and to record that in the child health record. If appropriate, this should ideally be in discussion with the father or primary care giver of the baby to ensure that they are aware of the need and benefit of newborn bloodspot screening.
- If the infant has been discharged home to the care of the father and the mother is too unwell to be discharged, the father should be instructed to bring the infant back into the hospital to obtain consent from the mother and then proceed to take the newborn bloodspot screening sample. This is similar to bringing infants back into hospital in areas where there is no weekend public health nursing service.
- Registered Public Health Nurses (RPHNs) arranging a house call to perform the newborn bloodspot screening must insist on the mother being present. Grandmothers or other relatives/friends cannot provide written consent.
- If a Midwife is taking the newborn bloodspot screening sample in hospital and the mother is not present on the ward, they should return when the mother is present.
- If there is social work involvement at the time of birth, the social worker should link with the Midwife or Public Health Nurse and the mother to ensure that

informed consent is obtained to perform the newborn bloodspot screening. However, in the absence of a full care order, only the parent(s)/legal guardian(s), or the mother if unmarried, can provide consent. An interim care order is not sufficient.

2.7.12.2 If the Parent(s)/legal Guardian(s) has literacy difficulties he/she can place a mark on the NBSC to indicate that they have been fully informed about the benefits and risks or newborn bloodspot screening.

2.7.12.3 The Sample Taker keeps the "Sample Takers" Copy of the NBSC and will file it in the CHR.

2.7.13 Parent(s)/Legal Guardian(s) right to opt out of NNBSP

2.7.13.1 The Parent(s)/legal Guardian(s) do have the right to opt-out from the NNBSP on behalf of their baby.

2.7.13.2 The Parent(s)/legal Guardian(s) should be discouraged from opting out in the best interest of their baby's health.

2.7.13.3 The Parent(s)/legal Guardian(s) will be fully informed of the potential consequences to their baby.

2.7.13.4 The Sample Taker informs the DPHN/designated Line Manager that the Parent(s)/legal Guardian(s) is opting out of the NNBSP and will discuss further actions required.

2.7.13.5 The Parent(s)/legal Guardian(s) must be requested to sign the *"HSE Opt-out Form"* (Appendix XIII). This form should be signed on the day of the Sample Takers visit. The Parent(s)/legal Guardian(s) can be given the opportunity to consult with other healthcare professionals if required.

2.7.13.6 All Parent(s)/legal Guardian(s) sign the *"HSE Opt-Out Form"* available at www.newbornscreening.ie in the presence of the Sample Taker and the Sample Taker signs the form.

- A copy of this form is given to the Parent(s)/legal Guardian(s).
- The original form is filed in the CHR.

2.7.13.7 The Sample Taker will send a copy of the *"HSE Opt-out Form"* to:

- The DPHN/CHO.
- The NNBSL.
- The baby's General Practitioner (GP).
- DOM/N/Maternity Hospital.

2.7.13.8 Where the Parent(s)/legal Guardian(s) have opted out of the programme, the Parent(s)/legal Guardian(s) will be informed that it is their responsibility to contact the Public Health Nursing Services and/or baby's GP should they change their mind in the future and wish to be included in the NNBSP. The RPHN/RM will record the discussion and decision made in the CHR.

2.7.13.9 Where Parent(s)/legal Guardian(s) decline to sign the *HSE Opt Out Form*:

- For Maternity Hospital /Unit, the sample taker will complete all the other steps, record the discussion with the Parent(s)/legal Guardian(s) and the decision made on the CHR. The sample taker will inform the baby's GP and RPHN.
- The RPHN/RM will complete all the other steps, record the discussion with the Parent(s)/legal Guardian(s) and the decision made on the CHR. The RPHN/RM will discuss this case with their Line Manager and decide on further actions.

2.7.14 Sample Collection

2.7.14.1 The Nurse/Midwife receives education and training on the procedure and can then carry out this procedure in compliance with their scope of practice (Scope of Nursing & Midwifery Practice Framework, NMBI 2015).

2.7.14.2 Registered Public Health Nurses (RPHNs), Registered Midwives (RMs) Registered Children's Nurses (RCNs), midwifery students, student PHNs, and Nursery Nurse (NN) undertake NBS under their scope of professional practice/competence:

- Where a RGN undertakes NBS, the DPHN and RGN must be satisfied that additional training has been undertaken and that the nurse is working within their scope of practice.
- Where a NN undertakes NBS, the DOMN/Hospital/Units and NN must be satisfied that additional training has been undertaken and that the NN is working within their scope of competence.

2.7.14.3 The method used for obtaining the blood sample is by "heel prick"

2.7.14.4 The blood sample should be taken **not earlier than 72 hours and not later than 120 hours** after the baby's birth and when feeding has been established. It is essential that all babies should receive an adequate protein intake before the sample is taken.

2.7.14.5 In view of the new conditions included in the newborn screening programme (particularly MCADD); there is no longer an emphasis on taking the sample at the end of the 72-120 hour window in breast fed babies. If protein intake is deemed to be suboptimal a further sample should be taken on or about Day 10 after birth. If protein intake is deemed to be suboptimal a further sample should be taken on or about Day 10 after birth.

2.7.14.6 Specific concerns for babies need to be considered and stated on the NBSC:

- **Total Parenteral Nutrition (TPN):** This should be clearly indicated on the NBSC because these babies may not be on any galactose containing feed and a Beutler test will need to be performed to rule-out Classical Galactosaemia.
- **Transfusion – Blood:** If a baby requires a red blood cell (RBC) transfusion before the routine 72-120 hour sample is taken, a pre RBC transfusion should be collected to perform a Beutler test to rule out Classical Galactosaemia. (Note: a RBC transfusion invalidates the Beutler test). The routine 72-120 hour sample should be collected as normal regardless if the child has a RBC transfusion and do not delay this 72-120 hour sample due to RBC transfusions. For any further samples to be collected on a RBC transfused baby then allow 72 hours to pass before taking any more samples.
- ***Prematurity:*** All premature babies should have the sample taken after 72 hours and before 120 hours from birth. Further samples should be collected at regular

intervals to a maximum of 4 samples until the baby is established on full feeds (for at least 24 hours) or at discharge.

- ***Prematurity:*** For very premature babies born before 30 weeks gestation the NBS should be repeated at discharge or at 36 weeks post conception age.
- **Meconium Ileus:** Cystic Fibrosis should be considered in those babies who present with meconium ileus within the first days of life. Send an ETDA sample to National Centre for Medical Genetics for CF mutation analysis, with full clinical information (CFTR mutational analysis is undertaken as Bloodspot IRT may give a false negative result in this clinical setting). A routine NBS sample should be taken at 72-120 hours to screen for the other conditions.
- **Maternal Phenylketonuria:** Phenylalanine is actively transported across the placenta; the blood level in the foetus is about twice that of the mother. Therefore, women who themselves have phenylketonuria, should plan conception, so that their condition is under optimum control at the time of conception. Regular and frequent monitoring of blood levels of phenylalanine and tyrosine are required throughout pregnancy in order to safeguard the well-being of the foetus. Following the birth of the baby a liquid sample should be taken from the baby at the same time as the NBS and a repeat sample at day 10 after birth.

2.7.14.7 Families who are deemed to be **high risk for any of the conditions being screened for,** require careful attention e.g. Traveller Families, or other Ethnic Groups, or a family history of any of the screened for conditions (Appendix XIV).

2.7.14.8 **The Beutler Test (For Classical Galactosaemia)** is completed for all babies born to Traveller parents and siblings of known cases of Classical Galactosaemia:

- A NBS sample should be taken from the baby immediately after the baby is born and prior to the first feed and/or before any blood transfusion has been given.
- The NBS sample is immediately sent to the NNBSL for a Beutler test – the NBSC should be clearly marked 'FOR BEUTLER TEST'. (Samples for Beutler must be in the laboratory by 10am on a Saturday).
- Analysis of Beutler samples and reporting of results can be obtained from the laboratory between 09:00-17:00 Monday to Friday and 09:00-12:00 Saturday. Bank Holiday opening hours of the NNBSL will be circulated in advance to DOM/Ns and DPHNs and posted on the website (Appendix XV)
- Following this NBS sample for the Beutler test, babies should be fed with a lactose/galactose free feed until the result of the Beutler Test result is known.
- These babies then will have the routine NBS sample taken between 72-120 hours.

2.7.14.9 Family history must be stated clearly on the NBSC and the condition indicated. High risk family history includes: siblings of known case of Phenylketonuria (PKU); Maple Syrup Urine Disease (MSUD); Homocystinuria (HCU); Classical Galactosaemia (Gal); Cystic Fibrosis (CF); Congenital Hypothyroidism (CHT); Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD); Glutaric Aciduria Type 1 (GA1) and cousins marriage in the Travelling Community.

2.7.15 Requirements for Sample Procedure

2.7.15.1 All sample takers must follow universal infection control standards while undertaking this sample

- Latex Free Gloves. Hand hygiene guidelines must be adhered to (Guidelines for Hand Hygiene in Irish Healthcare Setting, HCAI, 2015 and local policies).
- Sterile Haemolancet, controlled depth not more than 2.5 mm:
  - Generally 2.0 - 2.5mm is used for term babies.
  - For Preterm babies lancet depth maybe smaller and will depend on the prematurity of the baby.
- Sterile water (if required)
- Gauze
- Paper towel
- Sharps box
- Water Resistant Tear Proof Envelope (e.g. Tyvek or equivalent)
- Transport and drying box (for community services)
- Newborn Bloodspot Screening Card. Information must be correctly completed on the NBSC.
- Parent/Guardians Information Leaflet.

2.7.15.2 **The Sample Taker will** check the **expiry date** on the Card – this can be found on the bottom right hand corner under 'GPs Name'. The cards should not be used after this date. Using expired cards may result in a request for a repeat sample. Do not detach the bloodspot portion of the card from the main card, as it is bar-coded and is required for laboratory use.

2.7.15.3 **Sample takers need to note that:**

- Total Parental Nutrition should be clearly indicated on the NBSC; because these babies may not be on any galactose containing feed a Beutler test will need to be performed to rule-out Classical Galactosaemia.
- If the family have a history of PKU, HCU, MCADD and GA1, an additional NBSC is taken on day 10.

2.7.15.4 Unlabelled or inadequately labelled samples cannot be accepted for analysis.

2.7.15.5 **Biohazards** – babies whose mothers are known or suspected of being infected with HIV or Hepatitis B should have the screen performed. The NBSC must be identified as a Biohazard (See 2.7.17.7).

2.7.15.6 The sample taker working in CHO/LHO will require a water resistant and tear proof envelope (e.g. Tyvek) - and a Yellow Fluorescent address label (Regular Sample)[8].

2.7.15.7 NBSCs, appropriately packaged, should be sent daily and not batched. It is the responsibility of the sender to dispatch the sample as soon as possible after collection either by registered post or by courier.

2.7.15.8 **When a number of samples are being sent** in the one water resistant and tear proof envelope, ensure that all the samples are fully air dried and that the bloodspots are aligned at 180 degrees to each other so that the bloodspots do not rest on each other. Please ensure the checklist (Appendix XVI) is enclosed with:

[8] Each LHO prepare their own registered envelopes by obtaining, from their local major Post Office, a supply of the pre-paid plastic coated white A5 envelopes (Easipak pouches) and the blue 'Registered Post' bar-coded labels.

- The details of all the babies Surname and their UPIs
- The total number of NBSCs enclosed and the date posted
- For biohazards see 2.7.17.7.
- The name of the person compiling the checklist, their location and contact number
- Any additional comments

2.7.15.9 RPHN/RN/RGN notifies DPHN/designated officer that sample is taken.

2.7.16 Sample Technique

2.7.16.1In order to encourage blood flow to the heel area, the Parent(s)/Guardian(s) should be advised on the day prior to sampling (if possible) to put two pairs of socks on the baby's feet.

2.7.16.2 Do **NOT** touch bloodspot rings on the NBSC with gloves before, during or after the sample is taken. Ensure there is no contact with vaseline or other creams.

2.7.16.3 Latex interferes with Beutler sample and may cause a false positive result. Latex free gloves are to be used when carrying out the NBS.

2.7.16.4 Preferably take the sample from the baby while the Parent/legal Guardian cuddles the baby on his/her knee or breast feeds the baby. It also allows the Parent/legal Guardian to ask questions about the test. This can assist you but also comforts the baby.

2.7.16.5 Place a paper towel on the lap of the individual (RPHN/RM/Parent) holding the baby.

2.7.16.6 Ensure that the heel is visibly clean and warm**.** The skin may be gently rubbed for 1-2 minutes to increase blood supply if deemed necessary. Never use any external warming source (e.g. hairdryer, warm water). This introduces a risk of thermal burns and/or scalds.

2.7.16.7 Routine cleansing of the heel is not required. However, if the heel is visibly dirty or soiled it can be cleaned with gauze soaked in cool sterile water. Ensure that the heel is completely dry before taking the sample. Do not use alcohol or baby wipes as they may interfere with sample results or cause serum rings.

2.7.16.8 Skin to skin contact and allowing the baby's leg to hang lower than the body will encourage blood flow.

2.7.16.9 Encircle the heel with finger(s) and thumb and squeeze gently until the skin looks taut and suffused with blood.

2.7.16.10 Press the lancet firmly against the side of the ball of the heel and trigger the sterile lancet.

2.7.16.11 Hold the foot downwards and gently massage the heel to encourage blood flow.

2.7.16.12 Wipe away the first drop of blood and allow another large drop to form. (The first drop may be a diluted blood drop). Touch the circle marked on the card gently to the hanging drop so that the blood soaks through from the back of the card to the front:

- Blood drops must be soaked through from the back to the front of the card, filling all circles completely.
- Check that the blood has soaked completely through the circle on the front as well as the rear of the card.
- Do not press or squeeze the bloodspot to 'force' it through the NBSC.

2.7.16.13 Wipe away excess blood with gauze. Press clean gauze firmly onto wound until bleeding stops.

- It is not recommended that a plaster is used as this may be picked off by the baby and swallowed.

2.7.16.14 The Sample Taker will ensure sufficient blood is taken to meet the requirements to completely fill the four bloodspots circles on the NBSC.

2.7.16.15 See *Flowchart for Newborn Bloodspot Screening Sample Takers* if having difficulty in obtaining a sufficient sample – Appendix XVII

2.7.16.16 It is imperative that adequate blood is supplied for analysis to reduce the incidence of repeat sample requests. Please ensure that the blood soaks through fully from the back through to the front of the card to avoid an **insufficient** sample.

2.7.16.17 It is vitally important that all samples are **fully dried** before placing in the water resistant and tear proof envelope. **Wet samples** cause the production of 'serum rings' which could result in a false negative result and ultimately increases the risk of missing a case.

2.7.16.18 The sample taker will remove their gloves and wash their hands.

2.7.16.19 The sample taker in CHO/LHO will use a transport/drying box for the NBSC following the procedure to facilitate the transport of the NBSC from the baby's home to the Nurse's/Midwife's car in a safe manner (Appendix XVIII).

2.7.17 Transporting the NBSC to the NNBSL

2.7.17.1 The Sample Taker will forward the NBSC to the NNBSL. See Appendix XV for contact details for NNBSL.

2.7.17.2 If the NBS is taken in the Maternity Hospital/Unit the Nurse/Midwife/Nursery Nurse transfers the NBSC to the NNBSL through hospital arrangements for blood sampling products.

2.7.17.3 If the NBS is taken in the community, the RPHN/Nurse/Midwife in CHO/LHO will:

- Transfer the NBSC from the drying box and package according to the NNBSL regulations.
- A **water resistant and tear proof envelope** (e.g. Tyvek or equivalent) is used with the yellow fluorescent address label (Appendix XIX).
- It must be noted that a maximum of three NBSCs can be put into one water resistant and tear proof envelope. The samples must be fully air dried and assembled opposite

ends to each other i.e. to prevent blood circles from each card touching each other (See section 2.7.15.6 to 2.7.15.8).

2.7.17.4 Send the NBSC by **registered post**[9] (or by courier in exceptional circumstances i.e. emergency situations/extended holiday periods) to the NNBSL.

2.7.17.5 If more than a single NBSC is being dispatched together please include summary list of names/UPIs (Appendix XVI).

2.7.17.6 Dispose of lancet etc according to the local protocol for clinical waste – **do not include the lancet(s) in the package with the NBSC.**

2.7.17.7 **If a Biohazard e.g. HIV, Hepatitis A, B, C** is identified the NBSC must be managed in accordance with local infection control arrangement.

- The general principle is that the NBSC is placed in an inner sealed plastic envelope when the blood spot has completely dried.
  - The word 'Biohazard' may be noted on the NBSC, but the nature of the biohazard should not be noted as not relevant to screens being performed.
  - The Health Service Internal Biohazard label is placed on the inner sealed plastic envelope identifying a biohazard and alerting laboratory staff to this hazard. This separates the NBSC from other NBSC when a number of samples are being returned in the same **water resistant and tear proof envelope** (e.g. Tyvek (Appendix 12.19)).
  - This inner envelope is then placed in an **outer water resistant and tear proof envelope**. The outer envelope does not require the UN3373 sticker and 'Biohazard' does not need to be noted on the outer envelope.
- It is an offence to post a biohazard sample without its appropriate identification.
- It is also an offence to claim that a package is a biohazard when it is not.

2.7.17.8 For weekend/bank holiday and extended holiday periods the following procedures are applied:

- At weekends, where possible, samples taken on a Saturday are posted by 12 midday by registered post. Samples taken on Saturday afternoon/Sunday are posted on a Monday morning.
- At bank holiday weekends, where possible, samples taken on a Saturday are posted by 12midday by registered post. Samples taken on Saturday afternoon/Sunday/Monday are posted on a Tuesday morning.
- At extended holiday periods, a courier/taxi service may be used from a designated centre to the NNBSL for the NBSC to be transported directly to NNBSL.

2.7.18 Record Keeping

2.7.18.1The Midwife/Nurse/Nursery Nurse in the Maternity Hospital/Unit will record in the maternal healthcare record – child care section either:

- Completion of the procedure.
- Or issue a request for NBS to the relevant CHO/LHO or tertiary paediatric hospital.

[9] Receipt of posting is still required even if envelope is pre-paid. This is filed in the CHR.

2.7.18. The RPHN/Midwife/Nurse working in the **CHO/LHO** will record in the child health record the following:

- Confirmation of the NBS status (notification of receipt of request or confirmation of the date of sample taken in the maternity hospital/unit).
- Completion of the NBS procedure if it is taken in the community by filing the *Sample Taker* copy.
  - Confirmation to DPHN/designated officer, that the sample taking is completed.
  - Confirmation of postal registration: the number, date of sample taking and date of postage.
- Completion of the Primary Childhood Immunisation/GP Forename Card (White Card) (Appendix XX) if used and return it to the CHO/LHO Child Health Office where the information is used to verify notification of births inputted onto the Immunisation/PHR/Child Health Information ICT system and ensure a "fail safe" mechanism of checking that all babies are entered on the system. This Immunisation/PHR/Child Health Information ICT system is the full electronic birth register for the CHO/LHO. For some CHOs, fields from the system are extracted and used to populate the NBS register**. It is envisaged that the immunisation and child health databases will be integrated in future developments under NICHIS.**
- Receipt of registered post (available in Post Office at time of posting). *NOTE: All CHOs/LHOs have prepaid arrangements*.

2.7.18.3 The RPHN/Midwife/Nurse will check she/he has completed the sample collection correctly (Appendix XXI)

2.7.18.4 DPHNs and ADPHNs have access to the Immunisation /PHR /Child Health Information ICT System as a birth register. It is envisaged that PHNs will share this access in future developments under NICHIS.

2.7.19 Managing Inaccessible Visits

2.7.19.1 On failing to gain access to the Parent(s)/legal Guardian(s)'s home, the RPHN/RM/RN must phone the maternity hospital/unit to confirm the discharge details and discharge address of the mother and baby.

- Confirm the Parent/legal Guardian(s)'s Next of Kin with the Hospital.
- Telephone the Parent/legal Guardian(s)'s Next of Kin.
- If the next of kin is unsure of the Parent/legal Guardian(s)'s whereabouts, the RPHN/RM/RN inform them that further enquires will be made in order to establish the baby's whereabouts.
- Should the Next of Kin make contact with the Parent/legal Guardian(s)'s they are requested to ask the Parent(s)/ legal Guardian(s) to make contact with the RPHN/RM/RN.

2.7.19.2 If the above is not productive, the RPHN/RM/RN should phone the GP to see if there has been any recent contact with the Parent(s)/legal Guardian(s) and their baby and if he/she knows their whereabouts**.** The RPHN/RM/RN confirms the address and contact details, where possible.

2.7.19.3 All of the above steps should be documented, dated, timed and signed appropriately using the CHR.

2.7.19.4 The RPHN/RM/RN will inform the ADON/ADPHN throughout the above.

2.7.19.5 Following confirmation that it is the correct address and correct house the RPHN/RM/RN must leave a Business Card requesting the Parent(s)/legal Guardian(s) to return to the Maternity Hospital to have the test carried out.

2.7.19.6 In the event the address cannot be confirmed the RPHN/RM/RN does not leave a card but contacts the Maternity Hospital/Unit and/or GP for further details and records same in the CHR.

2.7.19.7 The RPHN/RM/RN contacts the Maternity Hospital/Unit with notice of the inaccessible visit and that the Parent(s)/legal Guardian(s) have been advised to return to the hospital for the NBST. The RPHN/RM/RN records this in the CHR.

2.7.19.8 The RPHN/RM/RN communicates with the Hospital/Unit the following day to ensure the Parent(s)/legal Guardian(s) has attended with the baby for the NBST

2.7.19.9 The Maternity Hospital /Unit documents that the NBST has been taken and informs the DPHN/Designated Officer in writing.

Stage Four: NNBSP Sample Results

2.7.20 NNBSL notify Maternity Hospital/Units/ DPHN, CHO of Sample Result

2.7.20.1 The NNBSL has an electronic reporting facility called eReports™ in use across the country since 2013/2014. Each Maternity Hospital and CHO/Local Health Office must nominate a NBS Management Lead and two authorised eReports™ users. This ensures eReports™ access is available at local level for authorised users.

2.7.20.2The NNBSL provides each authorised user with:

- User Name.
- User ID
- URL for eReports™ access.
- Handbook for eReports™ users
- Training as needed for authorised users.

2.7.20.3 Ereports™ allow:

- Verification of receipt of samples before results are available.
- Requests for repeat samples can be made electronically.
- Results of Samples become available 2-3 days after receipt of sample in the laboratory.
- Result codes are supplied which indicate interim stages of results analysis and investigations for some conditions.
- A search facility which can be used to find and link results, this includes searching by infant name, date of birth (DOB), or range of birth dates, mothers name, UPI etc.
- Results available online to authorised users in the CHO/LHO and the Hospitals/Units for up to 60 days following the issuing of a result.

- Individual reports can be printed locally for healthcare records and for parent/guardian information requests.

2.7.20.4 The NNBSL approves screening results normally within 48 hours of receipt of sample, which are made available via eReports™ to authorised users.

2.7.20.5 **The NNBSL phones the DOMN or designated officers and/or DPHN or designated officers with results requiring urgent action.**

2.7.20.6 The NNBSL forwards a list via eReports™ indicating the results of the screening to both:

- The DOMN/Maternity Hospital/Unit – designated officers.
- The CHO DPHN/LHOs – designated officers.
- Where relevant to the Specialist Children's Hospital.

Stage Five: Checking NNBSP Sample Results

2.7.21 Maternity Hospital/Units Checking NBS Register – sample results

2.7.21.1 The DOMN/designated officer (Outpatient Midwife Manager and/or Postnatal Midwife Manager) oversees and monitors NBS results for babies where a NBST has taken place in the Maternity Hospital/Unit.

2.7.21.2 The **Designated Officer** undertakes **daily** monitoring of the eReports™ for initial requests for NBS and for any repeat NBS requests and arranges appropriate sampling and resampling for any baby who remains in hospital at that point.

2.7.21.3 The **Designated Officer** undertakes **weekly monitoring procedures** as follows:

- Identifying and eliminating duplication.
- Identifying babies without UPI and ensuring all babies have a UPI issued.
- Monitoring on the newborn screening register those babies 12 days old where there are no eReport results identified. For those babies check the following:
  - That the sample has reached NNBSL (eReports™).
  - That the hospital/ward has a record of the NBSS been taken.
  - Or that a record of the "Opt Out" form (Appendix XIII) is in the maternal chart – baby section.
- Record closure of screening episode where information received identifies:
  - Confirmation that a baby has died.
  - The family have refused the service and an "Opt Out" form is completed (Appendix XIII).
- If sample has not been taken for other reasons, arrange sampling and continue to follow up until screening episode is closed.
- The designated officer signs/initials the entries to indicate that he/she has undertaken the exercise (note: manual format).

2.7.21.4 Conclusive results will normally be available within 18 days of birth for most conditions screened. Cystic Fibrosis results may not be available for up to 30 days after birth.

2.7.21.5 If results are not available by day eighteen, the designated officer should contact the NNBSL and request the result (Appendix XXII).

2.7.21.6 The designated officer will continue to follow up on results that are pending until all results are made available.

2.7.21.7 The Maternity Hospital/Unit is responsible for ensuring that omissions are notified to the DPHN/CHO where results have not been received from the NNBSL.

2.7.21.8 The Hospital /Designated Officer:

- Works with the CHO/LHO Child Health Lead, DPHN and/or their designated officers as required to ensure that all babies have a result recorded.
- The DPHN and their designated officers maintains an electronic/paper NBS register based on the full Birth Register (Immunisation/PHR/Child Health Information ICT System) and ensures all babies for the CHO/LHO have received the NNBSP and have a result recorded.

2.7.22 DPHN/CHO Checking NBS Register - sample results

2.7.22.1 DPHN Office/Designated Officer has direct access to the Immunisation/PHR/Child Health ICT system as an electronic full Birth Register for all babies in the CHO/LHO.

2.7.22.2 The DPHN Office/Designated Officer ensures homebirths and babies born outside the ROI are included in the Birth Register.

2.7.22.3 In CHO/LHOs which do not have capacity to include NBS results on the Immunisation/PHR/ Child Health ICT system, the data will be extracted from the Immunisation/PHR/Child Health ICT system to create a NBS Register (an Excel programme/ICT system). (It is envisaged that all CHO/LHOs will have capacity to record the NBS result on the Immunisation/PHR/Child Health ICT systems in future developments).

2.7.22.4 An excel spreadsheet/ICT system will have the fields as noted in Appendix X.

2.7.22.5 An excel spreadsheet/ICT system will readily facilitate counts from the requisite field as per Appendix X.

2.7.22.6 The DPHN/Designated Officer checks all NBS results against the Birth Register for the designated LHO area to ensure that **all babies** residing in the designated area have an NBS outcome status recorded by day 18.

2.7.22.7 The **Designated Officer** undertakes a **daily** monitoring of the eReports™ for initial requests for NBS and for any repeat NBS requests and informs the relevant ADPHN and/or PHN who arranges appropriate sampling and resampling for any baby who has returned home.

2.7.22.8 The **Designated Officer** undertakes "failsafe"[10] **weekly** monitoring procedures as follows:

[10] Failsafe means in this context a system designed to ensure that if one part of the system does not work, the whole system does not become dangerous.

- Confirmation that sample reached NNBSL through checking the eReports™ (i.e. check through "Patient Search" that the name of the baby is on eReports™ although results may not be available)
- If not reported within 4 days of sample collection (or by Day 12 if sample collection date is unknown), check that:
  - the Maternity Hospital/Unit of birth or SECM has completed the NBSS or
  - the RPHN/RM/RGN has taken the NBSS or
  - a record of the "Opt Out" form (Appendix XIII) is in the child health record or
  - baby is known to have "moved out" of the area or
  - baby has died.
- Record closure of screening episode where information received identifies:
  - Confirmation that a baby has died.
  - The family have refused the service and an "Opt Out Form" is completed (Appendix XIII).
  - The family moved to another area, which has taken over responsibility for NBSS.
- If the NBSS has not been taken for other reasons, arrange sampling and continue to follow up until screening episode is closed.
- Conclusive results will normally be available within 18 days of birth for most conditions screened. Cystic Fibrosis results may not be available for up to 30 days after birth.
- The designated officer signs/initials the entries to indicate that he/she has undertaken the exercise (Note: manual format).

2.7.22.9 Where confirmation is obtained through the eReports™ search that a sample has reached the NNBSL and no results or repeat request are received by Day 18, the designated officer makes a request for the results to the NNBSL (Appendix XXII).

2.7.22.10 Where reports are received which do not belong to the CHO/LHO, the designated officer will inform the NNBSL (Appendix XXIII).

2.7.22.11 The NBS Register should match the Immunisation/PHR/Child Health System. The designated officer will:

- Where babies have moved into the area and an eReport™ is received, the designated officer will inform the Immunisation/PHR/Child Health ICT system and add the baby to the NBS register.
- Where babies are on the NBS register which is aligned to the Immunisation/PHR/Child Health ICT system and an eReport™ is not received, the designated officer will contact the NNBSL (telephone or email) to ascertain is there a change of address or detail.
  - If the baby is residing in the area, the designated officer will request the result (Appendix XXII) and notify the Immunisation/PHR/Child Health ICT system of any change in details of the child.
  - If the baby is not residing in the area, the baby is ineligible for screening and the designated officer will notify the Immunisation/PHR/Child Health ICT system of any change in details of the child.

2.7.22.12 Where issues are identified by the designated officer in the monitoring procedures the designated officer will inform the Nurse Manager (DPHN/ADPHN) as appropriate.

2.7.22.13 The DPHN/designated officer informs the RPHN of the NBSS results**.** Individual results will be printed out from the eReport™ system (Appendix XXIV) and sent to the RPHN to be filed on the CHR.

2.7.22.14 The RPHN will file the sample results on the CHR and where results are pending the DPHN/designated officer monitors the results which are pending until confirmation of final results are received.

2.7.22.15 The RPHN will discuss the results with the Parent(s)/legal Guardian(s) at the next child health check (3 months PHN child health assessment).

2.7.22.16 Parent(s)/legal Guardian(s) can request a written copy of the sample result from the DPHN and /or NNBSL:

- Where the results are normal the DPHN can issue the results to the Parent(s)/legal Guardian(s).
- Where the results indicate an abnormal result, the DPHN will liaise with the NNSBL and/ or medical team before issuing the result to the Parent(s)/legal Guardian(s).

2.7.22.17 It is envisaged that the Public Health Nursing Services have access to Immunisation/PHR /Child Health ICT system as an electronic birth register and NBS register for all babies residing in their designated PCT Area. This will be considered as part of NICIS project.

Procedure for Repeat Newborn Bloodspot Screening Samples

Stage One: Management of positive results and repeat screening requests

2.7.23 Positive Results

2.7.23.1 The response to query positive result is immediate and direct.

2.7.23.2 The Liaison Nurse in the NNBSL contacts the designated liaison nurse in the maternity hospital/unit and:

- informs of the name, UPI, date of birth and address of the baby.
- the disorder suspected and the result of the screening test
- requests the liaison nurse in the maternity hospital/unit to locate the baby and Parent(s)/legal Guardian(s)

2.7.23.3 The Liaison Nurse in the NNBSL contacts the Parent(s)/legal Guardian(s) to:

- explain why the baby has to be referred to hospital
- explain what disorder is suspected in their baby, including the blood test result and any other test results such as the Beutler
- explain why a further blood sample is required
- arrange with the Parent(s)/ legal Guardian(s) for the baby to be brought directly to the Children's University Hospital, Temple Street or to the local Paediatric Unit as requested by the Newborn Bloodspot Screening Laboratory (Appendix XXV).
- advise the Parent(s)/legal Guardian(s) that their baby might be kept in hospital for a number of days depending on the result of the repeat investigation. Therefore they should bring a change of clothes for the baby and possibly for themselves.

2.7.23.4 The designated liaison nurse in the maternity hospital/unit may give the contact details of the Director of the National Newborn Bloodspot Screening Laboratory or deputy to parents if they wish to obtain more information before they arrive in the hospital.

2.7.23.5 Special arrangements have been put in place to contact Parent(s)/legal Guardian(s) with suspected CF where the Clinical Liaison Officer in the NNBSL will contact the CF Nurse Specialists in the appropriate HSE designated paediatric CF centres to give them the full contact details, relevant information and results of the mutational screen. The CF Nurse Specialist will book a sweat test appointment and then contact the Parent(s)/legal Guardian(s) to arrange for the baby to attend the appropriate CF centre.

2.7.23.6 At all times staff members must not instil any degree of anxiety or panic for the Parent(s)/legal Guardian(s), but must impart the information in a calm and professional manner, being fully informed of all the facts.

2.7.24 Repeat Screening Requests

2.7.24.1 Repeat samples are requested by the NNBSL where:

- Baby is too young when blood sample was collected.
- There are sample taker errors.
- There are borderline results and more definitive results are required.
- Insufficient blood on the NBSC for all tests to be performed.
- Unsatisfactory analysis: NBST needs to be rechecked (repeated); NBST difficult to interpret because specimen was contaminated or deteriorated during transit.
- There is a need for further definitive results to confirm conditions.
- Out of date cards used.
- Sample on the NBSC had not been dried properly before putting into the water and tear proof envelop causing “serum rings”.

2.7.24.2 The NNBSL will inform the DOMN/designated officer in hospital services and the DPHN/ designated officer in community services that a repeat NBSS is required.

2.7.24.3 The DOMN/Maternity Hospital/Unit and DPHN/CHO will establish the location of the baby:

- where the baby remains in hospital care, the hospital services will complete the request.
- where the baby has been discharged to Community Healthcare Organisations, the repeat sample will be completed by Public Health Nursing Service.

Stage Two: Repeat Sampling

2.7.25 Maternity Hospital/Unit repeat NNBS

2.7.25.1 The DOMN/Designated Officer in hospital services will ensure the request for the repeat NBSS is forwarded to the relevant ward on the day of receipt of request for repeat NBSS.

2.7.25.2 The RM/Nursery Nurse in the relevant ward will inform the Parent(s)/legal Guardian(s) of the reason for repeat sample taking.

2.7.25.3 Where the Parent(s)/legal Guardian(s) has been discharged from hospital care, the DPHN will be requested to complete the repeat NBSS.

2.7.25.4 Where the Parent(s)/legal Guardian(s) has been discharged from hospital care and if the DPHN office cannot be contacted within a timely timeframe, arrangements will be made for the Parent(s)/legal Guardian(s) and the baby to return to the hospital for sample taking.

2.7.26 Public Health Nursing Repeat NNBS

2.7.26.1 The DPHN/Designated Officer will ensure the request for the repeat NBSS is forwarded to the RPHN/RM/RGN on the day of receipt of request for repeat NBSS.

2.7.26.2 The RPHN/RM/RGN will contact the Parent(s)/legal Guardian(s) to arrange for sample taking either at a home visit or at a clinic visit within 24hours.

2.7.26.3 The repeat NBSS will be completed on the day of receipt of notification or the following day (within 24hours).

Stage Three: Procedure for Repeat Sampling

2.7.27 Repeat NBS Sampling

2.7.27.1 The sample taker will advise the Parent(s)/legal Guardian(s) about the reason for the repeat.

2.7.27.2 The sample taker must:

- explain to the Parent(s)/legal Guardian(s) why a repeat sample has been requested.
- assure the Parent(s)/legal Guardian(s) that if the repeat sample should prove positive, that they will be contacted immediately by a liaison nurse /Clinical Nurse Specialist from the relevant maternity hospital/unit programme.
- be aware that the Parent(s)/legal Guardian(s) can decide to opt out of the repeat sample being taken. Section 2.7.13 covers this in detail.

2.7.27.3 The sample will be taken as per Stage Two –Section 2.7.15-2.7.19.

2.7.27.4 The sample taker must clearly indicate on the NBSC that the sample is a Repeat Sample by ticking the box 'Yes' and if the repeat is requested for a specific condition as per the eReport™, then the sample taker should specify in the comments box on the NBSC what the repeat is for.

2.7.27.5 The sample should then be sent immediately to the NNBSL either by registered post or by courier or fast track services as appropriate.

2.7.27.6 The sample taker informs the DPHN and/or NNBSL where relevant that the sample is taken and gives details of how the sample is sent to the laboratory.

2.7.27.7 The sample taker will keep a record of the NBST in the baby section of the maternal chart or the neo-natal baby chart for hospital services and/or the Child Health Record for community services.

Stage Four: NBSP Repeat Sample Results

2.7.28 NNBSL notify Maternity Hospital/Unit/Community of repeat sample results

2.7.28.1 The NNBSL informs the DOMN/ Maternity Hospital/Unit and the DPHN/CHO of the repeat sample results (individual report) of the screening via eReports™.

2.7.28.2 The NNBSL follows up on all children where a result is not normal and/or requires clarification and follow up.

2.7.28.3 The NNBSL liaises with the designated liaison nurse in the relevant maternity hospital/unit who refers the baby and their Parent(s)/legal Guardian(s) to the appropriate care pathway if required.

2.7.28.4 The Medical Team/Clinical Nurse Specialist for the appropriate care pathway makes contact with the family and arranges a hospital /outpatient visit for babies with a suspected positive result (See section 2.7.23).

Stage Five: Checking NNBSP Repeat Sample Results

2.7.29 Checking NBSP register – sample results

2.7.29.1 The designated officer for the DOMN/Maternity Hospital/Unit and/or CHO DPHN for the designated LHO continues to follow up on repeat sample results until screening is closed.

PART B: PPPG Development Cycle

1.0 INITIATION

1.1 Purpose

To provide a standardised approach for the implementation of the National Newborn Bloodspot Screening Programme (NNBSP) in Maternity Hospitals/Units and Community Healthcare Organisations (CHOs) with their associated designated Local Health Offices (LHO).

1.2 Scope

The scope of this PPPG applies to:

1.2.1 The Director of Midwifery/Nursing (DOMN) and the management teams including the Midwifery Nursing Management Team within Maternity Hospitals/Units and designated staff who have responsibility for the NNBSP as follows

- Registered Midwives (RM)
- Registered Nurses (RN)
- Nursery Nurses
- Midwifery Students
- Administrative and clerical staff officers
- Consultant Neonatologist
- Consultant Paediatrician
- Non Consultant Hospital Doctors (NCHD)

1.2.2 The Director of Public Health Nursing (DPHN) and management teams within CHOs/LHOs in ensuring the implementation of the NNBSP within CHOs/LHOs

1.2.3 Registered Public Health Nurses (RPHNs), Registered Midwives, Self Employed Community Midwives (SECMs), Registered Nurses, Midwifery Students, Student PHNs working within CHOs/LHOs who are undertaking newborn bloodspot screening.

1.2.4 Designated Midwifery Officers and SECMs working under the direction of the HSE Memorandum of Understanding and Contractual Agreement (2014) or other private midwifery groups delivering midwifery services within the CHOs/LHOs, who undertake newborn bloodspot screening.

1.2.5 It must be noted that each Midwife/Nurse is accountable, both legally and professionally, for their own practice under the "*Scope of Nursing and Midwifery Practice Framework*" (Nursing and Midwifery Board of Ireland (NMBI) 2015) And "*The Code of Professional Conduct and Ethics for Registered Nurses and Registered Midwives*" (NMBI 2014).

1.2.6 Administrative staff members working within CHOs/LHOs supporting the role of the DPHN and Public Health Nursing service in providing the NNBSP.

1.2.7 Public Health Doctors, Community Medical Doctors and General Practitioners (GPs) within CHOs/LHOs to be aware of procedures for undertaking newborn bloodspot screening.

1.2.8 The NNBSP is offered to Parent(s)/Legal Guardian(s) of all babies who are born in Ireland, or who become Irish residents in the first year of life without having been screened for the conditions in the NNBSP in their country of origin. The conditions currently screened for in Ireland include:

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (HCU)

- Classical Galactosaemia (GAL)
- Cystic Fibrosis (CF)
- Congenital Hypothyroidism (CHT)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
- Glutaric Aciduria Type 1 (CA1)

Further detail on these eight conditions is contained in Appendix V.

1.3 Objective(s)

1.3.1 To ensure that the NNBSP is offered to all babies between 72 and 120 hours of birth and that all the screening test results follow the pathway and timeframe defined by the NNBSP Governance Group.

1.3.2 To guide all Registered Public Health Nurses (RPHNs), Registered Midwives (RMs), Self Employed Community Midwives (SECMs), Midwifery Groups and Registered Nurses (RNs), midwifery students, student PHNs, and Nursery Nurse staff members who are working in Hospitals/Units and Public Health Nursing Services within the CHOs/LHOs, supporting best practice in relation to the delivery of the NNBSP.

1.3.3 To guide administrative/clerical officers who have delegated roles from the Director of Midwifery/Nursing (DOMN) and Director of Public Health Nursing (DPHN) supporting the processes of the NNBSP in Maternity Hospitals/Units and Public Health Nursing services within CHOs/LHOs.

1.3.4 To guide Consultant Neonatologists, Consultant Paediatricians, Non Consultant Hospital Doctors (NCHD), Public Health Doctors, Community Medical Doctors and General Practitioners (GPs) who are working in Maternity Hospitals/Units and CHOs/LHOs supporting best practice in relation to the delivery of the NNBSP.

1.4 Outcome(s)

Parent(s)/Legal Guardian(s) of all babies who are born in Ireland, or who become Irish residents in the first year of life without having been screened for the conditions in the NNBSP in their country of origin, should be offered newborn bloodspot screening as per this procedure.

Any babies suspected of having any of the conditions screened for should be referred to the appropriate clinical care pathway.

1.5 PPPG Development Group

See Appendix II for Membership of the PPPG Development Group Template.
See Appendix III for PPPG Conflict of Interest Declaration Form Template.

1.6 PPPG Governance Group

1.6.1 See Appendix IV for Membership of the Approval Governance Group.

Note: This PPPG was approved in August 2016 by Primary Care and Health and Wellbeing Operations as per title page. There was a delay in implementation the PPPG due to a number of clarifications required due to changing organisational structures and personnel. There is a short revision date on this PPPG and an appropriate Approval Governance Group will be convened with the new structures at that time.

1.7 Supporting Evidence

1.7.1 **Relevant Legislation**

Notification of Births Act (1907)
Notification of Births Extension Act (1915)
Guardianship of the Infant Act (1964)
Health Act (1970)
Data Protection Acts (1988, 2003)
Child Care Act (1991)
Civil Registration Act (2004) and Civil Registration (Amendment Act (2014)
Nurse and Midwives Act (2011)
Child and Family Relationship Act (2015)

Relevant Guidelines/Policy

Best Health for Children (1999, revisited 2005)
Recording Clinical Practice Guidelines to Nurses and Midwives (NMBI, 2002)
Children First: National Guidance for the Protection and Welfare of Children (2011)
Standards and Recommended Practices for Healthcare Records (HSE, 2011)
National Standards for Safer Better Healthcare (HIQA, 2012)
The Code of Professional Conduct and Ethics for Registered Nurses and Registered Midwives (NMBI, 2014)
Practice Standards for Midwives (NMBI, 2015)
Scope of Nursing and Midwifery Practice Framework (NMBI, 2015)
National Infant Feeding Policy for Maternity and Neonatal services (HSE, 2015)
National Consent Policy (HSE, 2017)
Practical Guide to Newborn Bloodspot Screening in Ireland (7th Edition 2018)

1.7.2 This is the first national PPPG that has been developed in relation to the NNBSP. The Practical Guide to Newborn Bloodspot Screening, as developed by the National Newborn Bloodspot Screening Laboratory in Children's University Hospital Temple Street, has been the guidance document to date. This PPPG will operate in tandem with the Practical Guide and provide more detailed guidance for relevant staff in the Maternity Hospitals/Units and CHOs/LHOs.

1.7.3 List related PPPGs:
National Consent Policy (HSE, 2017)

1.8 Glossary of Terms (attach Appendix as appropriate).

ADPHN	Assistant Director of Public Health Nursing
ADON	Assistant Director of Nursing
CHR	Child Health Record
CF	Cystic Fibrosis
CGAL	Classical Galactosaemia
CHO	Community Healthcare Organisation
CHIS	Child Health Information System
CHT	Congenital Hypothyroidism
DPHN	Director of Public Health Nursing
DOMN	Director Of Midwifery and/or Nursing
GA1	Glutaric Aciduria Type 1
HA/HSA	Health Area/Health Service Area
HSE	Health Service Executive
HSCN	Health and Social Care Network
HIQA	Health Information and Quality Authority
HOD	Head of Discipline
HCU	Homocystinuria
IHI	Individual Health Identifier
LHO	Local Health Office
MCADD	Medium Chain Acyl-CoA Dehydrogenase Deficiency
MSUD	Maple Syrup Urine Disease
MN-CMS	Maternal and Newborn Clinical Management System
NBS	Newborn Bloodspot Screening
NBSC	Newborn Bloodspot Screening Card
NBSS	Newborn Bloodspot Screening Sample
NBST	Newborn Bloodspot Screening Test
NI	Northern Ireland
NICHIS	National Immunisation and Child Health Information System
NMBI	Nursing and Midwifery Board of Ireland
NN	Nursery Nurse
NNBSL	National Newborn Bloodspot Screening Laboratory
NNBSP	National Newborn Bloodspot Screening Programme
PCT	Primary Care Team
PCMT	Primary Care Management Team
PCUM	Primary Care Unit Management
PHNS	Public Health Nursing Services
PHR	Personal Health Record
PKU	Phenylketonuria
PPPGs	Policies, Procedures, Protocols and Guidelines
RM	Registered Midwife
RN	Registered Nurse (included General and/or Children's Nurses
ROI	Republic of Ireland
RPHN	Registered Public Health Nurse
SECM	Self Employed Community Midwife
SOP	Standard Operating Procedure
TOR	Terms of Reference
UPI	Unique Perinatal Identifier

2.0 DEVELOPMENT OF PPPG

2.1 List the questions (clinical/non-clinical)

What is newborn bloodspot screening?

Newborn bloodspot screening, also known as the 'heel-prick test' screens all newborn babies for eight rare conditions. A sample of blood is taken from the newborn baby's heel between 72 and 120 hours after birth and is sent to the National Newborn Bloodspot Screening Laboratory for analysis. The eight conditions screened for are:

- Phenylketonuria (PKU)
- Maple Syrup Urine Disease (MSUD)
- Homocystinuria (HCU)
- Classical Galactosaemia (GAL)
- Cystic Fibrosis (CF)
- Congenital Hypothyroidism (CHT)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
- Glutaric Aciduria Type 1 (GA1)

Why is it important?

Newborn bloodspot screening ensures that any babies with any these rare conditions are identified and treated as early as possible. All conditions that form part of the NNBSP have been selected because they all have a relatively high incidence within the Irish population and they fulfil, in part or in full, the criteria that have been set out internationally for newborn screening. These include:

- The conditions screened for are treatable
- There is a test available which is easily applied to large population groups
- There are few false positives and false negative results; i.e. the test is reliable
- The incidence of the conditions in the community is sufficiently high to warrant screening
- The cost of screening makes the process cost-effective

The incidence of the eight conditions screened for are noted below:

Condition	Date Started	Irish Incidence	Worldwide Incidence
Phenylketonuria	1966	1 : 4,500	1 : 12,000
Homocystinuria	1971	1 : 69,400	1 : 120,000
Classical Galactosaemia	1972	1: 16,200	1 : 45,000
Maple Syrup Urine Disease	1972	1 : 155,200	1 : 225,000
Congenital Hypothyroidism	1979	1 : 2,300	1 : 3,500
Cystic Fibrosis	2011	1 : 2,300	1 : 3,500
Medium Chain Acyl-CoA Dehydrogenase Deficiency	2018	1:66,000	1:14,600
Glutaric Aciduria type 1	2018	1:54,000	1:100,000

2.2 Describe the literature search strategy

The NNBSP has been operational since 1966. The NNBSL developed a "*Practical Guide to Newborn Bloodspot Screening in Ireland*" which formed the basis for how the programme operated. This procedure has been developed to compliment the '*Practical Guide'* and offer more detailed guidance for relevant staff. Please refer to the '*Practical Guide'* for more detail on the conditions, mode of inheritance etc.

2.3 Describe the method of appraising evidence

The NNBSP has been operational since 1966 and was one of the first national programmes in the world. The Irish programme has been tailored for the local population and the incidence of disorders occurring within the Irish population. The NNBSP will continue to develop as new information, treatments and evidence becomes available for these rare inherited disorders.

2.4 Describe the process the PPPG Development Group used to formulate recommendations

The NNBSP is already in operation using the "*Practical Guide to Newborn Bloodspot Screening in Ireland*" as a basis for its operation. This procedure has been developed to compliment the '*Practical Guide'* and offer more detailed guidance for relevant staff.

2.5 Provide a summary of the evidence from the literature

The NNBSP is already in operation using the "*Practical Guide to Newborn Bloodspot Screening in Ireland*" as a basis for its operation using best available evidence.

2.6 Detail resources necessary to implement the PPPG recommendations

The NNBSP is already in operation using the "*Practical Guide to Newborn Bloodspot Screening in Ireland*" as a basis for its operation. Resources are already in place.

2.7 Outline of PPPG steps/recommendations:

Procedure for taking initial newborn bloodspot screening sample

Stage One: Notification of birth to CHOs and requests for newborn bloodspot screening sample to Public Health Nursing services/LHOs

2.7.1 Notification of birth to CHO

2.7.1.1 Maternity Hospitals/Units, both within and outside of the Republic of Ireland (ROI), notify the CHO of all babies born and who reside in the designated CHO (Full Birth Notification – BNF-01). A copy of the BNF-01 is sent to:

- Registrar of births – demographic information
- Director of Public Health – clinical and demographic information
- National Perinatal Reporting System (NPRS) – clinical information
- Hospital/Unit copy – clinical and demographic information

It is noted that the copy for the Director of Public Health, in practice today, now goes to the CHO, community child health services/office or Director of Public Health Nursing Office.

2.7.1.2 The Maternity Hospital/Unit sends the Full Birth Notification to the local Child Health Office either electronically or in paper format[11]:

- For a number of CHOs (Dublin Areas), the Child Health Information Systems (CHIS) office receives the Full Birth Notification and inputs the information into the Child Immunisation/Parent Held Record (PHR) System. This information is electronically transferred on a daily basis to the DPHN Office in the LHO (12 midnight). The DPHN

[11] Birth Notifications should be made electronically as far as possible e.g. CHIS or secure email or Shared Drive systems and/or paper format. The practise of using fax should be minimised or discontinued.

Office/LHO set up a Child Health Record and forwards this to the Primary Care Team (PCT) RPHN.

- For other CHOs, the Full Birth Notification information is received at one or many locations, the DPHN Office, the Birth Notification Office, the Child Health Office and/or Primary Care Unit Management (PCUM)[12]. By agreement, the DPHN Office and Birth Notification/PCUM, input the information into the Child Immunisation/Child Health/PHR System. The RPHNs set up a Child Health Record within local guidelines which includes newborn screening.
- The development and roll out of the Maternal and Newborn Clinical Management system (MN-CMS) and National Immunisation and Child Health Immunisation System (NICHIS) will standardise the collection of data further.

2.7.2 Maternity Hospital/Unit Births and NNBSP

2.7.2.1 The Maternity Hospital/Unit is responsible for completing the newborn bloodspot screening test (NBST) for all babies on the wards between 72 and 120 hours following the birth and delegating duties to the relevant Midwives/Nurses. Where babies are being discharged before 72 hours, the Maternity Hospital/Unit is responsible for notifying the relevant CHO/designated LHO Public Health Nursing Service of the requirement to carry out the NBST.

2.7.2.2 The Maternity Hospital/Unit:

- Maintains a birth register for all babies born in the hospital/unit.
- Issues the **Unique Perinatal Identifier (UPI)** number for all babies born in the Hospital/Unit (Appendix VI). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- Maintains a NBS Register for babies who have NBS completed while in Hospital.
- Delegates duties for maintaining the NBS Register to a designated officer.

2.7.2.3 The Maternity Hospital/Unit forwards the NBSC taken within the requisite timeframe (72-120 hours) to the NNBSL and confirms to the DPHN the date and time NBS sample was obtained from the baby.

2.7.2.4 If the sample is not taken within the Hospital before discharge, the DOMN/designated staff member is responsible for ensuring that:

- The designated officer in the CHO/LHO is notified of the birth.
- The DPHN[13] /Child Health Office[14] is informed that the baby has been discharged prior to the sample being carried out and is notified of the request for NBS[15]. The Hospital/Unit should provide two contact details of Parent(s)/legal Guardian(s) including mobile telephone number if known. Where a Parent(s)/legal Guardian(s) and baby are staying at a temporary address this should be made clear on the NBS notification.

[12] It is recommended that all child health databases and birth notifications be coordinated and managed by a Child Health Office for the CHO/LHO.

[13] For the purposes of this document the point of contact between Hospital and Primary Care for NBS is the office of the DPHN. Some CHOs have a Liaison PHN acting on behalf of the DPHN, coordinating birth notifications and NBS requests, screening discharges from hospitals and maintaining birth and NBS registers for the CHO/LHO for audit and collation of key performance indicators.

[14] Some CHOs/LHOs the child health office/immunisation/central referrals office coordinates birth notifications and NBS requests on behalf of the office of the DPHN.

[15] NBS notifications should be made electronically as far as possible e.g. secure email or shared drive systems and/or paper format. The practice of using fax should be minimised or discontinued.

2.7.2.5 The Maternity Ward within the Maternity/Unit Hospital has responsibility for:

- Completing an **Early Discharge Summary/Edited Birth Notification** for all babies that require the NBS sample to be taken in the CHO/LHO and forwarding it to the designated DPHN.
- The date the newborn bloodspot screening sample (NBSS) is required to be taken should be clearly written on the Discharge Summary/Edited Birth Notification.
- Completing the following information on the NBSC: The **Baby's UPI number; Time of Birth; Date of first feed**; **Type of Feed.** The mother will be given the *HSE Parent/Guardian Information Leaflet* (Appendix VII).
- Notifying the DPHN if the child or mother is **infected with a pathogen** e.g. HIV or Hepatitis B**,** which would result in the card not being exempt from the **Biohazard packaging regulations**.
- Giving the mother the **Discharge Checklist: Baby.** The mother will give this discharge summary to the PCT RPHN (Appendix VIII).

2.7.2.6 In the case of early discharges or where the Maternity Hospital/Unit wish to observe the baby, the DOMN/designated staff member may issue an appointment for the baby to return to the hospital for follow up during the designated timeframe for which the NBS can be undertaken.

2.7.2.7 The Maternity Hospital/Unit may request that mother and baby return to the Hospital/Unit at weekends/bank holidays/extended holiday periods, to facilitate early discharge and where the Public Health Nursing Service does not provide a weekend service.

2.7.3 Hospital /Unit Transfer to Tertiary Hospital

2.7.3.1 It may be necessary for a baby to be transferred to a tertiary hospital for continuing medical and/or surgical treatment before the NBSS has been taken.

2.7.3.2 The nurse/midwife responsible for the transfer of a baby from a maternity unit/hospital to a tertiary referral paediatric unit must inform the receiving unit that the NBST is required and give them the baby's UPI.

2.7.3.3 The receiving paediatric unit must have written procedures for:

- Performing the test between 72 and 120 hours after birth.
- Sending the newborn bloodspot screening sample (NBSS) to the NNBSL.
- Recording the results in the baby's medical records.
- Informing the Maternity Hospital/Unit of the NBST results.

2.7.4 Hospital/Unit Notification of NBS Request to PHN Service

2.7.4.1 Maternity Hospital/Unit, within and outside the ROI, notify the request to carry out NBS to the Public Health Nursing Service for the designated LHO. This may be part of the pending discharge (Discharge Summary/Edited Birth Notification and/or Telephone contact/Fax)[16].

- In the case of a family and baby relocating and/or returning to reside in ROI (from outside ROI) prior to the NBST, it is the responsibility of the Maternity Hospital/Unit to inform the DPHN for the designated LHO that the baby and family is relocating/returning within the ROI, within the timeframe for the sample to be taken (72-120 hours).

[16] NBS Notifications should be made electronically as far as possible e.g. Secure email or Shared Drive systems, and/or paper format. The practice of using fax should be minimised or discontinued.

- It is the responsibility of the Maternity Hospital/Unit to inform the relevant Public Health Nursing Service/LHO in the case of a family and baby discharging to a temporary address (e.g. grandmother) prior to returning to the address of their main residence.
- Maternity Hospitals/Units can use the Health Atlas address finder facility to help check the correct LHO to write on the NBSC. https://finder.healthatlasireland.ie/
- Maternity Hospitals/Units are reminded that it is important for the screening process that both temporary and permanent addresses are flagged on the NBSC (on separate sheet if appropriate) and the 36 hour birth notification to the LHO is completed, in order to ensure that immediate follow up is facilitated and to clarify where baby will reside on a permanent basis and avail of other child health services.

2.7.4.2 Where a request for NBS comes from a Maternity Hospital/Unit to an incorrect LHO, the DPHN/LHO must return the NBS request to the Maternity Hospital/Unit. The Maternity Hospital/Unit must correct this information and send the request to the correct LHO and notify the NNBSL of the change of LHO.

2.7.4.3 Northern Ireland (NI) Maternity Hospitals/Units will contact the Public Health Nursing Service/LHO and request:

- The Public Health Nursing Service Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.4.4 Please note that in the case of babies born outside the jurisdiction, the Parent(s)/legal Guardian(s) are equally responsible for ensuring that the baby is screened. If the Parent(s)/legal Guardian(s) notify the Public Health Nursing Service of the birth on their return to this jurisdiction, the Public Health Nursing Service is responsible for ensuring that the sample is carried out in this jurisdiction. The RPHN/RM may seek the advice of the NNBSL and/or Family GP.

2.7.5 Self Employed Community Midwife (SECM) notification of birth and NBS status to PHN service

2.7.5.1 The SECM is responsible for undertaking NBS for all babies in their care who require the NBST. The NBS sample will be taken between 72 hours and 120 hours following the birth.

2.7.5.2 The SECM will contact the DPHN/LHO and request:

- The Public Health Nursing Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.5.3 The SECM will notify the DPHN for the designated LHO of the home birth and pending discharge from SCEM service (Discharge Summary/Edited Birth Notification).

2.7.6 Midwifery Groups notification of NBS status to PHN service

2.7.6.1 Midwifery Groups contracted by the Parent(s)/legal Guardian(s) to provide postnatal care are responsible for undertaking NBS for all babies in their care who require the NBST. The NBS sample will be taken between 72 and 120 hours following the birth.

2.7.6.2 Midwifery Groups will contact the DPHN for the designated LHO and request:

- The DPHN to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that the UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL.

2.7.6.3 The Midwifery Group will notify the DPHN for the designated LHO that the NBS sample has been taken and the pending discharge from the Midwifery groups service.

2.7.7 General Practitioner notification of the NBS status to PHN service

2.7.7.1 In the event that a GP undertakes the NBST, they must ensure the NBS sample is taken between 72 hours and 120 hours following the birth.

2.7.7.2 The GP will contact the DPHN/LHO and request:

- The Public Health Nursing Office/Child Health Office to issue the Baby's **Unique Perinatal Identifier** (UPI) number (Appendix IX). It is noted that UPI will be replaced by the Individual Health Identifier (IHI) when implemented.
- This UPI will be inserted on the NBSC prior to forwarding the NBSC to the NNBSL

2.7.7.3 The GP will notify the DPHN for the designated LHO that the NBST has been taken.

2.7.7.4 In the case of Traveller babies, the baby may have moved out of the area before the NBST has been carried out. The DPHN must be informed of the circumstances as early as possible so that alternative arrangements may be made.

2.7.8 Self-referral to PHN service and NBS status

2.7.8.1 Where a Parent(s)/legal Guardian(s) presents to the Public Health Nursing Service and notification of the baby's birth and requirement for newborn bloodspot screening has not been communicated from hospital services, the Public Health Nursing Service will provide the required service. This situation may arise for babies born outside of the ROI.

Stage Two: DPHN Office and Notification to designated PCT PHN

2.7.9 Notification to the PCT PHN

2.7.9.1 The DPHN/Designated Officer will

- Ensure on receipt of request for NBS via **Early Discharge Summary/Edited Birth Notification** and/or telephone contact, that the relevant details of the request are correct, including the UPI number for the baby (Appendix VI).
- Ensure that the full details of the baby are correct, including details of the mother and baby who have an infection such as HIV or Hepatitis B that requires specific consideration (See 2.7.15.5 and2.7.17.7) are highlighted to the practitioner.
- When a NBS request is for an incorrect LHO, the DPHN/Designated Officer will notify the Maternity Hospital/Unit of the error immediately.

- Issue a UPI number to Hospital/Unit and SECM, Midwifery Group on request for babies born outside of ROI and home births (Appendix IX).
- Have access to a full electronic Birth Register. The Immunisation/PHR/Child Health Information System is the electronic birth register for the LHO. The Child Health Office coordinates all immunisation and child health databases.
- In the short term, for some LHOs, "fields" from the Immunisation System/PHR/Child Health Information System are extracted and used to populate the NBS Register. For some LHOs, the Immunisation System/PHR/Child Health Information System is the birth register and the NBS register as it has the facility to enter the required data.

2.7.9.2 The DPHN or Designated Officer will:

- Receive the data extract from the Immunisation/PHR/Child Health Information System.
- Input the additional information on the NBS Register. (Appendix X)
- Notify the designated Primary Care Team (PCT) Registered Public Health Nurse (RPHN)/Registered Midwife (RM)/Registered Nurse (RGN) of the request for the NBS by email or fax.

2.7.9.3 The email will generate an automatic acknowledgement of receipt of notification. Where email is not used the RPHN/RM/RGN acknowledges receipt of the request for Newborn Bloodspot Screening via telephone or fax[17].

2.7.9.4 During the normal working week, the RPHN/RM/RGN contacts the Parent(s)/legal Guardian(s) and arranges an agreed time and place for NBS to be carried out. This may be a:

- Home visit.
- Centre based visit if this is more convenient for the Parent(s)/legal Guardian(s) and/or RPHN/RM/RGN.

2.7.9.5 At weekends and bank holiday weekends:

- For some LHOs, where there is a weekend/bank holiday period service for newborn babies, the Designated Officer informs the weekend Nurse by 12:00 midday on Friday that a NBSS is requested for the baby.
- For some LHOs, where there is no Public Health Nursing Service weekend/bank holiday period service, the Maternity Hospital /Unit requests that the mother, with her baby, return to the Hospital/Unit. The NBST can be performed either in the postnatal ward or at a planned NBS clinic. The Public Health Nursing Service will be informed by the Hospital/Unit of the birth and that the NBSS was taken, on the following Monday/Tuesday after weekend/bank holiday period.
- For some LHOs, where the Maternity Hospital/Unit provides early discharge/domino schemes, the Hospital/Unit requests that the mother, with her baby, return to the Hospital/Unit and/or Community Midwife for the NBS to be undertaken. The NBSS can be performed either in the postnatal ward or at a planned NBS clinic. The Public Health Nursing Service will be informed of the birth and that the NBSS was taken on the following Monday/Tuesday after weekend/bank holiday period.
- As per local arrangements, additional special arrangements can be organised for extended holiday periods/ urgent requests from NNBSL, to ensure the NBSS is delivered from LHO to the NNBSL (For example a courier arrangement).

[17] The practise of using fax should be minimised or discontinued.

2.7.9.6 Where a family and baby are resident temporarily in the LHO immediately following birth (e.g. with Grandmother) evidence of newborn bloodspot screening must be obtained. This may take the form of email or written results or a written record in the Child Health Record (CHR). Screening must be carried out if the baby has not been screened. Please note the baby will need to be added to the relevant Child Health Database (Immunisation/PHR/NBS Register).

2.7.9.7 Where a baby under one year of age has moved into the LHO (temporary or permanent), evidence of NBS must be obtained. This may take the form of email or written results or a written record in the CHR. Where no proof of testing is available it should be assumed that the baby is untested. Screening a child who has not been screened, testing should be discussed with the NNBSL, and where relevant the GP, together with the parent. If recommended for testing, screening must be arranged. In general, all conditions are eligible for testing in unscreened children under one year except Cystic Fibrosis where the screening test is invalid from 6 weeks after birth.

2.7.9.8 Where a baby, up to one year of age, enters the country and has not received NBS for all the conditions screened in the Irish NNBSP in the baby's country of origin, clarification will be sought from the NNBSL and the baby's family GP regarding the required screening for the baby. In general, all of the conditions (except Cystic Fibrosis, where the screening test is invalid from 6 weeks after birth) should be screened for in unscreened children under one year.

2.7.9.9 The decision to screen children greater than one year is a local decision, made by the clinician and family GP and is dependent on the family history and the country of origin of the parents. In such circumstances the NNBSL should be contacted for advice in advance of sample taking.

2.7.9.10 Red Weather alerts: In the event of a red weather alert due to adverse weather conditions, the sample taker in conjunction with the ADOM/ADPHN must assess the risk of travel of either the PHN or the parent and baby against the risk of delayed screening. Further advice can be sought from the laboratory on such occasions.

Stage Three: Procedure for NBS sample taking

2.7.10 Information Giving

2.7.10.1 Parent(s)/legal Guardian(s) should have received information on the NNBSP antenatally in the 3rd trimester and at discharge from hospital/units.

2.7.10.2 All Parent(s)/legal Guardian(s) will be given:

- The NNBSP *HSE Parent/Guardian Information Leaflet* (Appendix VII) if they have not already received this information. Information Leaflets can be obtained on the website: *www.newbornscreening.ie* including information leaflets in a selection of other languages.
- The Parent Information Copy (Appendix XI) of the *Newborn Bloodspot Screening Card* (NBSC) (Appendix XII) which contains information about the NNBSP.

2.7.10.3 All Parent(s)/legal Guardian(s) will receive the following information:

- The conditions which are being screened for and the reason for screening.
- Advise that further samples may be required.
- The NBSC will be retained for 10 years (Note: NBSCs area currently retained indefinitely pending decision by the Minister of Health).

2.7.10.4 The Parent who signs the NBSC must be the legal Guardian. Please note:

- If the Parents are married, both Parents are legal Guardians and either can sign the NBSC.
- If Parents are not married, the mother is automatically the legal Guardian of the baby and can sign the NBSC. The unmarried father may not be automatically the guardian of the baby, and may not be in a position within the timeframe for the NBS to have legal responsibility to sign the NBSC.

2.7.11 Completing the Newborn Screening Card

2.7.11.1 The Sample Taker completes the details required for the NBSC:

- *Baby's Unique Perinatal Identifier (*UPI)
- *Babies demographic details:* Baby's surname, first name, address including postal code/eircode.
- *Place of birth: Hospital or Community.*
- *Babies healthcare record number/transfer to another hospital number*
- *Mother's surname*
- *Local Health Office*
- *Location sample taken:* Please tick the relevant box 'Community' or 'Hospital'.
- If the NBSS is taken in a hospital, other than the birth hospital, please enter the hospital in which the sample is actually taken and the specific Children's/General Hospital/Department/Ward.
- *GP's Name.*
- *Gestational age; time of birth; date of birth, gender.*
- Birth Weight; Rank (For multiple births e.g. Twin 1 Twin 2); date of first feed.
- *Blood Transfusion*; Date and time of first transfusion, date and time of last second transfusion.
- *Type of Feed:* breast, artificial, Total Parenteral Nutrition (TPN), IV fluids, soya/lactose free. Some sample-takers have ticked the incorrect box for **Type of Feed.** The box on the **LEFT** of the **title** of the feed must be ticked.
- *Comments: Family History, Beutler, Meconium Ileus.*
- *Date and time of collection, repeat specimen (yes or no).*
- *Sample taker name and phone contact number:* This requires the phone number of the **sample taker** (or of other individual/centre as determined locally by the DPHN) to enable the sample taker to be contacted directly should this be required. The Parent(s)/legal Guardian(s) phone number is **NOT** to be entered here.
- *Parent(s)/legal Guardian(s) Preferred language:* This field should always be completed – for English, or any other language, as an interpreter could be required to communicate possible positive results and the need to attend a hospital for further samples.
- *Parent(s)/legal Guardian(s) consent signature.*
- *Please note details on the reverse side of the NBSC will be filled in by the laboratory.*

2.7.11.2 The Sample Taker when completing the NBSC ensures that:

- The top information sheet is removed and given to the Parent(s)/legal Guardian(s) together with the parent copy (2nd sheet of NBSC) at the time of sampling.
- Parent(s)/legal Guardian(s) will be informed that results will be issued to the maternity hospitals/units and LHOs only. Results will not be issued to Parent(s)/legal Guardian(s) over the telephone.
- Further information is available on www.newbornscreening.ie or by e-mailing info.newbornscreening@cuh.ie

2.7.11.3 Where there is an error in recording the 'type of feed' identified, the NNBSL needs to be notified immediately by emailing info.newbornscreening@cuh.ie identifying the service location, stating the baby's clinical details and the amendment to be made.

2.7.12 Consent

2.7.12.1 The Parent(s)/legal Guardian(s) by signing the NBSC confirm that:

- He/she received, read and understood the *HSE Parent/Guardian Information Leaflet.*
- The details of the baby on the NBSC are correct.
- They consent to the screening blood sample being taken and a repeat sample if required
- They agree to the storage of the NBSC as per current Department of Health recommendations.

Who can give consent?

- Married parents – if the mother and father are married at the time of birth then either can give consent to screening as they are joint guardians of the infant as per Section 6 of the *Guardianship of Infants Act 1964*
- Unmarried parents – if the mother and father are unmarried at the time of birth only the mother can give consent as per Section 6(4)of the *Guardianship of Infants Act 1964*
- If the mother is unavailable to sign the consent, i.e. through illness or hospital transfer, the unmarried father cannot sign the consent. In these cases, the sample taker should make every effort to contact the mother to get verbal consent and to document this in the relevant clinical notes/child health record. This may include liaising with the mother's medical team to obtain developments on her condition and position to provide consent for the newborn bloodspot screening sample to be taken.
- If the mother is not contactable, for example due to severe inpatient medical illness, then the HSE must act in the best interest of the infant which would be to take the newborn bloodspot screening sample and inform the mother as soon as possible as to the decision taken and to record that in the child health record. If appropriate, this should ideally be in discussion with the father or primary care giver of the baby to ensure that they are aware of the need and benefit of newborn bloodspot screening.
- If the infant has been discharged home to the care of the father and the mother is too unwell to be discharged, the father should be instructed to bring the infant back into the hospital to obtain consent from the mother and then proceed to take the newborn bloodspot screening sample. This is similar to bringing infants back into hospital in areas where there is no weekend public health nursing service.
- Registered Public Health Nurses (RPHNs) arranging a house call to perform the newborn bloodspot screening must insist on the mother being present.

Grandmothers or other relatives/friends cannot provide written consent.

- If a Midwife is taking the newborn bloodspot screening sample in hospital and the mother is not present on the ward, they should return when the mother is present.
- If there is social work involvement at the time of birth, the social worker should link with the Midwife or Public Health Nurse and the mother to ensure that informed consent is obtained to perform the newborn bloodspot screening. However, in the absence of a full care order, only the parent(s)/legal guardian(s), or the mother if unmarried, can provide consent. An interim care order is not sufficient.

2.7.12.2 If the Parent(s)/legal Guardian(s) has literacy difficulties he/she can place a mark on the NBSC to indicate that they have been fully informed about the benefits and risks or newborn bloodspot screening.

2.7.12.3 The Sample Taker keeps the "Sample Takers" Copy of the NBSC and will file it in the CHR.

2.7.13 Parent(s)/Legal Guardian(s) right to opt out of NNBSP

2.7.13.1 The Parent(s)/legal Guardian(s) do have the right to opt-out from the NNBSP on behalf of their baby.

2.7.13.2 The Parent(s)/legal Guardian(s) should be discouraged from opting out in the best interest of their baby's health.

2.7.13.3 The Parent(s)/legal Guardian(s) will be fully informed of the potential consequences to their baby.

2.7.13.4 The Sample Taker informs the DPHN/designated Line Manager that the Parent(s)/legal Guardian(s) is opting out of the NNBSP and will discuss further actions required.

2.7.13.5 The Parent(s)/legal Guardian(s) must be requested to sign the *"HSE Opt-out Form"* (Appendix XIII). This form should be signed on the day of the Sample Takers visit. The Parent(s)/legal Guardian(s) can be given the opportunity to consult with other healthcare professionals if required.

2.7.13.6 All Parent(s)/legal Guardian(s) sign the *"HSE Opt-Out Form"* available at www.newbornscreening.ie in the presence of the Sample Taker and the Sample Taker signs the form.

- A copy of this form is given to the Parent(s)/legal Guardian(s).
- The original form is filed in the CHR.

2.7.13.7 The Sample Taker will send a copy of the *"HSE Opt-out Form"* to:

- The DPHN/CHO.
- The NNBSL.
- The baby's General Practitioner (GP).
- DOM/N/Maternity Hospital.

2.7.13.8 Where the Parent(s)/legal Guardian(s) have opted out of the programme, the Parent(s)/legal Guardian(s) will be informed that it is their responsibility to contact the Public Health Nursing Services and/or baby's GP should they change their mind in the future and wish to be included in the NNBSP. The RPHN/RM will record the discussion and decision made in the CHR.

2.7.13.9 Where Parent(s)/legal Guardian(s) decline to sign the *HSE Opt Out Form*:

- For Maternity Hospital /Unit, the sample taker will complete all the other steps, record the discussion with the Parent(s)/legal Guardian(s) and the decision made on the CHR. The sample taker will inform the baby's GP and RPHN.
- The RPHN/RM will complete all the other steps, record the discussion with the Parent(s)/legal Guardian(s) and the decision made on the CHR. The RPHN/RM will discuss this case with their Line Manager and decide on further actions.

2.7.14 Sample Collection

2.7.14.1 The Nurse/Midwife receives education and training on the procedure and can then carry out this procedure in compliance with their scope of practice (Scope of Nursing & Midwifery Practice Framework, NMBI 2015).

2.7.14.2 Registered Public Health Nurses (RPHNs), Registered Midwives (RMs) Registered Children's Nurses (RCNs), midwifery students, student PHNs, and Nursery Nurse (NN) undertake NBS under their scope of professional practice/competence:

- Where a RGN undertakes NBS, the DPHN and RGN must be satisfied that additional training has been undertaken and that the nurse is working within their scope of practice.
- Where a NN undertakes NBS, the DOMN/Hospital/Units and NN must be satisfied that additional training has been undertaken and that the NN is working within their scope of competence.

2.7.14.3 The method used for obtaining the blood sample is by "heel prick"

2.7.14.4 The blood sample should be taken **not earlier than 72 hours and not later than 120 hours** after the baby's birth and when feeding has been established. It is essential that all babies should receive an adequate protein intake before the sample is taken.

2.7.14.5 In view of the new conditions included in the newborn screening programme (particularly MCADD); there is no longer an emphasis on taking the sample at the end of the 72-120 hour window in breast fed babies. If protein intake is deemed to be suboptimal a further sample should be taken on or about Day 10 after birth. If protein intake is deemed to be suboptimal a further sample should be taken on or about Day 10 after birth.

2.7.14.6 Specific concerns for babies need to be considered and stated on the NBSC:

- **Total Parenteral Nutrition (TPN):** This should be clearly indicated on the NBSC because these babies may not be on any galactose containing feed and a Beutler test will need to be performed to rule-out Classical Galactosaemia.
- **Transfusion – Blood:** If a baby requires a red blood cell (RBC) transfusion before the routine 72-120 hour sample is taken, a pre RBC transfusion should be collected to perform a Beutler test to rule out Classical Galactosaemia. (Note: a RBC

transfusion invalidates the Beutler test). The routine 72-120 hour sample should be collected as normal regardless if the child has a RBC transfusion and do not delay this 72-120 hour sample due to RBC transfusions. For any further samples to be collected on a RBC transfused baby then allow 72 hours to pass before taking any more samples.

- ***Prematurity:*** All premature babies should have the sample taken after 72 hours and before 120 hours from birth. Further samples should be collected at regular intervals to a maximum of 4 samples until the baby is established on full feeds (for at least 24 hours) or at discharge.
- ***Prematurity:*** For very premature babies born before 30 weeks gestation the NBS should be repeated at discharge or at 36 weeks post conception age.
- **Meconium Ileus:** Cystic Fibrosis should be considered in those babies who present with meconium ileus within the first days of life. Send an ETDA sample to National Centre for Medical Genetics for CF mutation analysis, with full clinical information (CFTR mutational analysis is undertaken as Bloodspot IRT may give a false negative result in this clinical setting). A routine NBS sample should be taken at 72-120 hours to screen for the other conditions.
- **Maternal Phenylketonuria:** Phenylalanine is actively transported across the placenta; the blood level in the foetus is about twice that of the mother. Therefore, women who themselves have phenylketonuria, should plan conception, so that their condition is under optimum control at the time of conception. Regular and frequent monitoring of blood levels of phenylalanine and tyrosine are required throughout pregnancy in order to safeguard the well-being of the foetus. Following the birth of the baby a liquid sample should be taken from the baby at the same time as the NBS and a repeat sample at day 10 after birth.

2.7.14.7 Families who are deemed to be **high risk for any of the conditions being screened for,** require careful attention e.g. Traveller Families, or other Ethnic Groups, or a family history of any of the screened for conditions (Appendix XIV).

2.7.14.8 **The Beutler Test (For Classical Galactosaemia)** is completed for all babies born to Traveller parents and siblings of known cases of Classical Galactosaemia:

- A NBS sample should be taken from the baby immediately after the baby is born and prior to the first feed and/or before any blood transfusion has been given.
- The NBS sample is immediately sent to the NNBSL for a Beutler test – the NBSC should be clearly marked 'FOR BEUTLER TEST'. (Samples for Beutler must be in the laboratory by 10am on a Saturday).
- Analysis of Beutler samples and reporting of results can be obtained from the laboratory between 09:00-17:00 Monday to Friday and 09:00-12:00 Saturday. Bank Holiday opening hours of the NNBSL will be circulated in advance to DOM/Ns and DPHNs and posted on the website (Appendix XV)
- Following this NBS sample for the Beutler test, babies should be fed with a lactose/galactose free feed until the result of the Beutler Test result is known.
- These babies then will have the routine NBS sample taken between 72-120 hours.

2.7.14.9 Family history must be stated clearly on the NBSC and the condition indicated. High risk family history includes: siblings of known case of Phenylketonuria (PKU); Maple Syrup Urine Disease (MSUD); Homocystinuria (HCU); Classical Galactosaemia (Gal); Cystic Fibrosis (CF);

Congenital Hypothyroidism (CHT); Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCADD); Glutaric Aciduria Type 1 (GA1) and cousins marriage in the Travelling Community.

2.7.15 Requirements for Sample Procedure

2.7.15.1 All sample takers must follow universal infection control standards while undertaking this sample

- Latex Free Gloves. Hand hygiene guidelines must be adhered to (Guidelines for Hand Hygiene in Irish Healthcare Setting, HCAI, 2015 and local policies).
- Sterile Haemolancet, controlled depth not more than 2.5 mm:
  - Generally 2.0 - 2.5mm is used for term babies.
  - For Preterm babies lancet depth maybe smaller and will depend on the prematurity of the baby.
- Sterile water (if required)
- Gauze
- Paper towel
- Sharps box
- Water Resistant Tear Proof Envelope (e.g. Tyvek or equivalent)
- Transport and drying box (for community services)
- Newborn Bloodspot Screening Card. Information must be correctly completed on the NBSC.
- Parent/Guardians Information Leaflet.

2.7.15.2 **The Sample Taker will** check the **expiry date** on the Card – this can be found on the bottom right hand corner under 'GPs Name'. The cards should not be used after this date. Using expired cards may result in a request for a repeat sample. Do not detach the bloodspot portion of the card from the main card, as it is bar-coded and is required for laboratory use.

2.7.15.3 **Sample takers need to note that:**

- Total Parental Nutrition should be clearly indicated on the NBSC; because these babies may not be on any galactose containing feed a Beutler test will need to be performed to rule-out Classical Galactosaemia.
- If the family have a history of PKU, HCU, MCADD and GA1, an additional NBSC is taken on day 10.

2.7.15.4 Unlabelled or inadequately labelled samples cannot be accepted for analysis.

2.7.15.5 **Biohazards** – babies whose mothers are known or suspected of being infected with HIV or Hepatitis B should have the screen performed. The NBSC must be identified as a Biohazard (See 2.7.17.7).

2.7.15.6 The sample taker working in CHO/LHO will require a water resistant and tear proof envelope (e.g. Tyvek) - and a Yellow Fluorescent address label (Regular Sample)[18].

[18] Each LHO prepare their own registered envelopes by obtaining, from their local major Post Office, a supply of the pre-paid plastic coated white A5 envelopes (Easipak pouches) and the blue 'Registered Post' bar-coded labels.

2.7.15.7NBSCs, appropriately packaged, should be sent daily and not batched. It is the responsibility of the sender to dispatch the sample as soon as possible after collection either by registered post or by courier.

2.7.15.8 **When a number of samples are being sent** in the one water resistant and tear proof envelope, ensure that all the samples are fully air dried and that the bloodspots are aligned at 180 degrees to each other so that the bloodspots do not rest on each other. Please ensure the checklist (Appendix XVI) is enclosed with:

- The details of all the babies Surname and their UPIs
- The total number of NBSCs enclosed and the date posted
- For biohazards see 2.7.17.7.
- The name of the person compiling the checklist, their location and contact number
- Any additional comments

2.7.15.9 RPHN/RN/RGN notifies DPHN/designated officer that sample is taken.

2.7.16 Sample Technique

2.7.16.1In order to encourage blood flow to the heel area, the Parent(s)/Guardian(s) should be advised on the day prior to sampling (if possible) to put two pairs of socks on the baby's feet.

2.7.16.2 Do **NOT** touch bloodspot rings on the NBSC with gloves before, during or after the sample is taken. Ensure there is no contact with vaseline or other creams.

2.7.16.3 Latex interferes with Beutler sample and may cause a false positive result. Latex free gloves are to be used when carrying out the NBS.

2.7.16.4 Preferably take the sample from the baby while the Parent/legal Guardian cuddles the baby on his/her knee or breast feeds the baby. It also allows the Parent/legal Guardian to ask questions about the test. This can assist you but also comforts the baby.

2.7.16.5 Place a paper towel on the lap of the individual (RPHN/RM/Parent) holding the baby.

2.7.16.6 Ensure that the heel is visibly clean and warm**.** The skin may be gently rubbed for 1-2 minutes to increase blood supply if deemed necessary. Never use any external warming source (e.g. hairdryer, warm water). This introduces a risk of thermal burns and/or scalds.

2.7.16.7 Routine cleansing of the heel is not required. However, if the heel is visibly dirty or soiled it can be cleaned with gauze soaked in cool sterile water. Ensure that the heel is completely dry before taking the sample. Do not use alcohol or baby wipes as they may interfere with sample results or cause serum rings.

2.7.16.8 Skin to skin contact and allowing the baby's leg to hang lower than the body will encourage blood flow.

2.7.16.9 Encircle the heel with finger(s) and thumb and squeeze gently until the skin looks taut and suffused with blood.

2.7.16.10 Press the lancet firmly against the side of the ball of the heel and trigger the sterile lancet.

2.7.16.11 Hold the foot downwards and gently massage the heel to encourage blood flow.

2.7.16.12 Wipe away the first drop of blood and allow another large drop to form. (The first drop may be a diluted blood drop). Touch the circle marked on the card gently to the hanging drop so that the blood soaks through from the back of the card to the front:

- Blood drops must be soaked through from the back to the front of the card, filling all circles completely.
- Check that the blood has soaked completely through the circle on the front as well as the rear of the card.
- Do not press or squeeze the bloodspot to 'force' it through the NBSC.

2.7.16.13 Wipe away excess blood with gauze. Press clean gauze firmly onto wound until bleeding stops.

- It is not recommended that a plaster is used as this may be picked off by the baby and swallowed.

2.7.16.14 The Sample Taker will ensure sufficient blood is taken to meet the requirements to completely fill the four bloodspots circles on the NBSC.

2.7.16.15 See *Flowchart for Newborn Bloodspot Screening Sample Takers* if having difficulty in obtaining a sufficient sample – Appendix XVII

2.7.16.16 It is imperative that adequate blood is supplied for analysis to reduce the incidence of repeat sample requests. Please ensure that the blood soaks through fully from the back through to the front of the card to avoid an **insufficient** sample.

2.7.16.17 It is vitally important that all samples are **fully dried** before placing in the water resistant and tear proof envelope. **Wet samples** cause the production of 'serum rings' which could result in a false negative result and ultimately increases the risk of missing a case.

2.7.16.18 The sample taker will remove their gloves and wash their hands.

2.7.16.19 The sample taker in CHO/LHO will use a transport/drying box for the NBSC following the procedure to facilitate the transport of the NBSC from the baby's home to the Nurse's/Midwife's car in a safe manner (Appendix XVIII).

2.7.17 Transporting the NBSC to the NNBSL

2.7.17.1 The Sample Taker will forward the NBSC to the NNBSL. See Appendix XV for contact details for NNBSL.

2.7.17.2 If the NBS is taken in the Maternity Hospital/Unit the Nurse/Midwife/Nursery Nurse transfers the NBSC to the NNBSL through hospital arrangements for blood sampling products.

2.7.17.3 If the NBS is taken in the community, the RPHN/Nurse/Midwife in CHO/LHO will:

- Transfer the NBSC from the drying box and package according to the NNBSL regulations.
- A **water resistant and tear proof envelope** (e.g. Tyvek or equivalent) is used with the yellow fluorescent address label (Appendix XIX).
- It must be noted that a maximum of three NBSCs can be put into one water resistant and tear proof envelope. The samples must be fully air dried and assembled opposite ends to each other i.e. to prevent blood circles from each card touching each other (See section 2.7.15.6 to 2.7.15.8).

2.7.17.4 Send the NBSC by **registered post**[19] (or by courier in exceptional circumstances i.e. emergency situations/extended holiday periods) to the NNBSL.

2.7.17.5 If more than a single NBSC is being dispatched together please include summary list of names/UPIs (Appendix XVI).

2.7.17.6 Dispose of lancet etc according to the local protocol for clinical waste – **do not include the lancet(s) in the package with the NBSC.**

2.7.17.7 **If a Biohazard e.g. HIV, Hepatitis A, B, C** is identified the NBSC must be managed in accordance with local infection control arrangement.

- The general principle is that the NBSC is placed in an inner sealed plastic envelope when the blood spot has completely dried.
  - The word 'Biohazard' may be noted on the NBSC, but the nature of the biohazard should not be noted as not relevant to screens being performed.
  - The Health Service Internal Biohazard label is placed on the inner sealed plastic envelope identifying a biohazard and alerting laboratory staff to this hazard. This separates the NBSC from other NBSC when a number of samples are being returned in the same **water resistant and tear proof envelope** (e.g. Tyvek (Appendix 12.19)).
  - This inner envelope is then placed in an **outer water resistant and tear proof envelope**. The outer envelope does not require the UN3373 sticker and 'Biohazard' does not need to be noted on the outer envelope.
- It is an offence to post a biohazard sample without its appropriate identification.
- It is also an offence to claim that a package is a biohazard when it is not.

2.7.17.8 For weekend/bank holiday and extended holiday periods the following procedures are applied:

- At weekends, where possible, samples taken on a Saturday are posted by 12 midday by registered post. Samples taken on Saturday afternoon/Sunday are posted on a Monday morning.
- At bank holiday weekends, where possible, samples taken on a Saturday are posted by 12midday by registered post. Samples taken on Saturday afternoon/Sunday/Monday are posted on a Tuesday morning.
- At extended holiday periods, a courier/taxi service may be used from a designated centre to the NNBSL for the NBSC to be transported directly to NNBSL.

[19] Receipt of posting is still required even if envelope is pre-paid. This is filed in the CHR.

2.7.18 Record Keeping

2.7.18.1 The Midwife/Nurse/Nursery Nurse in the Maternity Hospital/Unit will record in the maternal healthcare record – child care section either:

- Completion of the procedure.
- Or issue a request for NBS to the relevant CHO/LHO or tertiary paediatric hospital.

2.7.18. The RPHN/Midwife/Nurse working in the **CHO/LHO** will record in the child health record the following:

- Confirmation of the NBS status (notification of receipt of request or confirmation of the date of sample taken in the maternity hospital/unit).
- Completion of the NBS procedure if it is taken in the community by filing the *Sample Taker* copy.
- Confirmation to DPHN/designated officer, that the sample taking is completed.
- Confirmation of postal registration: the number, date of sample taking and date of postage.
- Completion of the Primary Childhood Immunisation/GP Forename Card (White Card) (Appendix XX) if used and return it to the CHO/LHO Child Health Office where the information is used to verify notification of births inputted onto the Immunisation/PHR/Child Health Information ICT system and ensure a "fail safe" mechanism of checking that all babies are entered on the system. This Immunisation/PHR/Child Health Information ICT system is the full electronic birth register for the CHO/LHO. For some CHOs, fields from the system are extracted and used to populate the NBS register**. It is envisaged that the immunisation and child health databases will be integrated in future developments under NICHIS.**
- Receipt of registered post (available in Post Office at time of posting). *NOTE: All CHOs/LHOs have prepaid arrangements*.

2.7.18.3 The RPHN/Midwife/Nurse will check she/he has completed the sample collection correctly (Appendix XXI)

2.7.18.4 DPHNs and ADPHNs have access to the Immunisation /PHR /Child Health Information ICT System as a birth register. It is envisaged that PHNs will share this access in future developments under NICHIS.

2.7.19 Managing Inaccessible Visits

2.7.19.1 On failing to gain access to the Parent(s)/legal Guardian(s)'s home, the RPHN/RM/RN must phone the maternity hospital/unit to confirm the discharge details and discharge address of the mother and baby.

- Confirm the Parent/legal Guardian(s)'s Next of Kin with the Hospital.
- Telephone the Parent/legal Guardian(s)'s Next of Kin.
- If the next of kin is unsure of the Parent/legal Guardian(s)'s whereabouts, the RPHN/RM/RN inform them that further enquires will be made in order to establish the baby's whereabouts.
- Should the Next of Kin make contact with the Parent/legal Guardian(s)'s they are requested to ask the Parent(s)/ legal Guardian(s) to make contact with the RPHN/RM/RN.

2.7.19.2 If the above is not productive, the RPHN/RM/RN should phone the GP to see if there has been any recent contact with the Parent(s)/legal Guardian(s) and their baby and if he/she knows their whereabouts**.** The RPHN/RM/RN confirms the address and contact details, where possible.

2.7.19.3 All of the above steps should be documented, dated, timed and signed appropriately using the CHR.

2.7.19.4 The RPHN/RM/RN will inform the ADON/ADPHN throughout the above.

2.7.19.5 Following confirmation that it is the correct address and correct house the RPHN/RM/RN must leave a Business Card requesting the Parent(s)/legal Guardian(s) to return to the Maternity Hospital to have the test carried out.

2.7.19.6 In the event the address cannot be confirmed the RPHN/RM/RN does not leave a card but contacts the Maternity Hospital/Unit and/or GP for further details and records same in the CHR.

2.7.19.7 The RPHN/RM/RN contacts the Maternity Hospital/Unit with notice of the inaccessible visit and that the Parent(s)/legal Guardian(s) have been advised to return to the hospital for the NBST. The RPHN/RM/RN records this in the CHR.

2.7.19.8 The RPHN/RM/RN communicates with the Hospital/Unit the following day to ensure the Parent(s)/legal Guardian(s) has attended with the baby for the NBST

2.7.19.9 The Maternity Hospital /Unit documents that the NBST has been taken and informs the DPHN/Designated Officer in writing.

Stage Four: NNBSP Sample Results

2.7.20 NNBSL notify Maternity Hospital/Units/ DPHN, CHO of Sample Result

2.7.20.1 The NNBSL has an electronic reporting facility called eReports™ in use across the country since 2013/2014. Each Maternity Hospital and CHO/Local Health Office must nominate a NBS Management Lead and two authorised eReports™ users. This ensures eReports™ access is available at local level for authorised users.

2.7.20.2The NNBSL provides each authorised user with:

- User Name.
- User ID
- URL for eReports™ access.
- Handbook for eReports™ users
- Training as needed for authorised users.

2.7.20.3 Ereports™ allow:

- Verification of receipt of samples before results are available.
- Requests for repeat samples can be made electronically.
- Results of Samples become available 2-3 days after receipt of sample in the laboratory.

- Result codes are supplied which indicate interim stages of results analysis and investigations for some conditions.
- A search facility which can be used to find and link results, this includes searching by infant name, date of birth (DOB), or range of birth dates, mothers name, UPI etc.
- Results available online to authorised users in the CHO/LHO and the Hospitals/Units for up to 60 days following the issuing of a result.
- Individual reports can be printed locally for healthcare records and for parent/guardian information requests.

2.7.20.4 The NNBSL approves screening results normally within 48 hours of receipt of sample, which are made available via eReports™ to authorised users.

2.7.20.5 **The NNBSL phones the DOMN or designated officers and/or DPHN or designated officers with results requiring urgent action.**

2.7.20.6 The NNBSL forwards a list via eReports™ indicating the results of the screening to both:

- The DOMN/Maternity Hospital/Unit – designated officers.
- The CHO DPHN/LHOs – designated officers.
- Where relevant to the Specialist Children's Hospital.

Stage Five: Checking NNBSP Sample Results

2.7.21 Maternity Hospital/Units Checking NBS Register – sample results

2.7.21.1 The DOMN/designated officer (Outpatient Midwife Manager and/or Postnatal Midwife Manager) oversees and monitors NBS results for babies where a NBST has taken place in the Maternity Hospital/Unit.

2.7.21.2 The **Designated Officer** undertakes **daily** monitoring of the eReports™ for initial requests for NBS and for any repeat NBS requests and arranges appropriate sampling and resampling for any baby who remains in hospital at that point.

2.7.21.3 The **Designated Officer** undertakes **weekly monitoring procedures** as follows:

- Identifying and eliminating duplication.
- Identifying babies without UPI and ensuring all babies have a UPI issued.
- Monitoring on the newborn screening register those babies 12 days old where there are no eReport results identified. For those babies check the following:
  - That the sample has reached NNBSL (eReports™).
  - That the hospital/ward has a record of the NBSS been taken.
  - Or that a record of the "Opt Out" form (Appendix XIII) is in the maternal chart – baby section.
- Record closure of screening episode where information received identifies:
  - Confirmation that a baby has died.
  - The family have refused the service and an "Opt Out" form is completed (Appendix XIII).
- If sample has not been taken for other reasons, arrange sampling and continue to follow up until screening episode is closed.

- The designated officer signs/initials the entries to indicate that he/she has undertaken the exercise (note: manual format).

2.7.21.4 Conclusive results will normally be available within 18 days of birth for most conditions screened. Cystic Fibrosis results may not be available for up to 30 days after birth.

2.7.21.5 If results are not available by day eighteen, the designated officer should contact the NNBSL and request the result (Appendix XXII).

2.7.21.6 The designated officer will continue to follow up on results that are pending until all results are made available.

2.7.21.7 The Maternity Hospital/Unit is responsible for ensuring that omissions are notified to the DPHN/CHO where results have not been received from the NNBSL.

2.7.21.8 The Hospital /Designated Officer:

- Works with the CHO/LHO Child Health Lead, DPHN and/or their designated officers as required to ensure that all babies have a result recorded.
- The DPHN and their designated officers maintains an electronic/paper NBS register based on the full Birth Register (Immunisation/PHR/Child Health Information ICT System) and ensures all babies for the CHO/LHO have received the NNBSP and have a result recorded.

2.7.22 DPHN/CHO Checking NBS Register - sample results

2.7.22.1 DPHN Office/Designated Officer has direct access to the Immunisation/PHR/Child Health ICT system as an electronic full Birth Register for all babies in the CHO/LHO.

2.7.22.2 The DPHN Office/Designated Officer ensures homebirths and babies born outside the ROI are included in the Birth Register.

2.7.22.3 In CHO/LHOs which do not have capacity to include NBS results on the Immunisation/PHR/ Child Health ICT system, the data will be extracted from the Immunisation/PHR/Child Health ICT system to create a NBS Register (an Excel programme/ICT system). (It is envisaged that all CHO/LHOs will have capacity to record the NBS result on the Immunisation/PHR/Child Health ICT systems in future developments).

2.7.22.4 An excel spreadsheet/ICT system will have the fields as noted in Appendix X.

2.7.22.5 An excel spreadsheet/ICT system will readily facilitate counts from the requisite field as per Appendix X.

2.7.22.6 The DPHN/Designated Officer checks all NBS results against the Birth Register for the designated LHO area to ensure that **all babies** residing in the designated area have an NBS outcome status recorded by day 18.

2.7.22.7 The **Designated Officer** undertakes a **daily** monitoring of the eReports™ for initial requests for NBS and for any repeat NBS requests and informs the relevant ADPHN and/or PHN who arranges appropriate sampling and resampling for any baby who has returned home.

2.7.22.8 The **Designated Officer** undertakes "failsafe"[20] **weekly** monitoring procedures as follows:

- Confirmation that sample reached NNBSL through checking the eReports™ (i.e. check through "Patient Search" that the name of the baby is on eReports™ although results may not be available)
- If not reported within 4 days of sample collection (or by Day 12 if sample collection date is unknown), check that:
  - the Maternity Hospital/Unit of birth or SECM has completed the NBSS or
  - the RPHN/RM/RGN has taken the NBSS or
  - a record of the "Opt Out" form (Appendix XIII) is in the child health record or
  - baby is known to have "moved out" of the area or
  - baby has died.
- Record closure of screening episode where information received identifies:
  - Confirmation that a baby has died.
  - The family have refused the service and an "Opt Out Form" is completed (Appendix XIII).
  - The family moved to another area, which has taken over responsibility for NBSS.
- If the NBSS has not been taken for other reasons, arrange sampling and continue to follow up until screening episode is closed.
- Conclusive results will normally be available within 18 days of birth for most conditions screened. Cystic Fibrosis results may not be available for up to 30 days after birth.
- The designated officer signs/initials the entries to indicate that he/she has undertaken the exercise (Note: manual format).

2.7.22.9 Where confirmation is obtained through the eReports™ search that a sample has reached the NNBSL and no results or repeat request are received by Day 18, the designated officer makes a request for the results to the NNBSL (Appendix XXII).

2.7.22.10 Where reports are received which do not belong to the CHO/LHO, the designated officer will inform the NNBSL (Appendix XXIII).

2.7.22.11 The NBS Register should match the Immunisation/PHR/Child Health System. The designated officer will:

- Where babies have moved into the area and an eReport™ is received, the designated officer will inform the Immunisation/PHR/Child Health ICT system and add the baby to the NBS register.
- Where babies are on the NBS register which is aligned to the Immunisation/PHR/Child Health ICT system and an eReport™ is not received, the designated officer will contact the NNBSL (telephone or email) to ascertain is there a change of address or detail.
  - If the baby is residing in the area, the designated officer will request the result (Appendix XXII) and notify the Immunisation/PHR/Child Health ICT system of any change in details of the child.

[20] Failsafe means in this context a system designed to ensure that if one part of the system does not work, the whole system does not become dangerous.

- If the baby is not residing in the area, the baby is ineligible for screening and the designated officer will notify the Immunisation/PHR/Child Health ICT system of any change in details of the child.

2.7.22.12 Where issues are identified by the designated officer in the monitoring procedures the designated officer will inform the Nurse Manager (DPHN/ADPHN) as appropriate.

2.7.22.13 The DPHN/designated officer informs the RPHN of the NBSS results**.** Individual results will be printed out from the eReport™ system (Appendix XXIV) and sent to the RPHN to be filed on the CHR.

2.7.22.14 The RPHN will file the sample results on the CHR and where results are pending the DPHN/designated officer monitors the results which are pending until confirmation of final results are received.

2.7.22.15 The RPHN will discuss the results with the Parent(s)/legal Guardian(s) at the next child health check (3 months PHN child health assessment).

2.7.22.16 Parent(s)/legal Guardian(s) can request a written copy of the sample result from the DPHN and /or NNBSL:

- Where the results are normal the DPHN can issue the results to the Parent(s)/legal Guardian(s).
- Where the results indicate an abnormal result, the DPHN will liaise with the NNSBL and/ or medical team before issuing the result to the Parent(s)/legal Guardian(s).

2.7.22.17 It is envisaged that the Public Health Nursing Services have access to Immunisation/PHR /Child Health ICT system as an electronic birth register and NBS register for all babies residing in their designated PCT Area. This will be considered as part of NICIS project.

Procedure for Repeat Newborn Bloodspot Screening Samples

Stage One: Management of positive results and repeat screening requests

2.7.23 Positive Results

2.7.23.2 The response to query positive result is immediate and direct.

2.7.23.2 The Liaison Nurse in the NNBSL contacts the designated liaison nurse in the maternity hospital/unit and:

- informs of the name, UPI, date of birth and address of the baby.
- the disorder suspected and the result of the screening test
- requests the liaison nurse in the maternity hospital/unit to locate the baby and Parent(s)/legal Guardian(s)

2.7.23.3 The Liaison Nurse in the NNBSL contacts the Parent(s)/legal Guardian(s) to:

- explain why the baby has to be referred to hospital
- explain what disorder is suspected in their baby, including the blood test result and any other test results such as the Beutler

- explain why a further blood sample is required
- arrange with the Parent(s)/ legal Guardian(s) for the baby to be brought directly to the Children's University Hospital, Temple Street or to the local Paediatric Unit as requested by the Newborn Bloodspot Screening Laboratory (Appendix XXV).
- advise the Parent(s)/legal Guardian(s) that their baby might be kept in hospital for a number of days depending on the result of the repeat investigation. Therefore they should bring a change of clothes for the baby and possibly for themselves.

2.7.23.4 The designated liaison nurse in the maternity hospital/unit may give the contact details of the Director of the National Newborn Bloodspot Screening Laboratory or deputy to parents if they wish to obtain more information before they arrive in the hospital.

2.7.23.5 Special arrangements have been put in place to contact Parent(s)/legal Guardian(s) with suspected CF where the Clinical Liaison Officer in the NNBSL will contact the CF Nurse Specialists in the appropriate HSE designated paediatric CF centres to give them the full contact details, relevant information and results of the mutational screen. The CF Nurse Specialist will book a sweat test appointment and then contact the Parent(s)/legal Guardian(s) to arrange for the baby to attend the appropriate CF centre.

2.7.23.6 At all times staff members must not instil any degree of anxiety or panic for the Parent(s)/legal Guardian(s), but must impart the information in a calm and professional manner, being fully informed of all the facts.

2.7.24 Repeat Screening Requests

2.7.24.1 Repeat samples are requested by the NNBSL where:

- Baby is too young when blood sample was collected.
- There are sample taker errors.
- There are borderline results and more definitive results are required.
- Insufficient blood on the NBSC for all tests to be performed.
- Unsatisfactory analysis: NBST needs to be rechecked (repeated); NBST difficult to interpret because specimen was contaminated or deteriorated during transit.
- There is a need for further definitive results to confirm conditions.
- Out of date cards used.
- Sample on the NBSC had not been dried properly before putting into the water and tear proof envelop causing "serum rings".

2.7.24.2 The NNBSL will inform the DOMN/designated officer in hospital services and the DPHN/ designated officer in community services that a repeat NBSS is required.

2.7.24.3 The DOMN/Maternity Hospital/Unit and DPHN/CHO will establish the location of the baby:

- where the baby remains in hospital care, the hospital services will complete the request.
- where the baby has been discharged to Community Healthcare Organisations, the repeat sample will be completed by Public Health Nursing Service.

Stage Two: Repeat Sampling

2.7.25 Maternity Hospital/Unit repeat NNBS

2.7.25.1 The DOMN/Designated Officer in hospital services will ensure the request for the repeat NBSS is forwarded to the relevant ward on the day of receipt of request for repeat NBSS.

2.7.25.2 The RM/Nursery Nurse in the relevant ward will inform the Parent(s)/legal Guardian(s) of the reason for repeat sample taking.

2.7.25.3 Where the Parent(s)/legal Guardian(s) has been discharged from hospital care, the DPHN will be requested to complete the repeat NBSS.

2.7.25.4 Where the Parent(s)/legal Guardian(s) has been discharged from hospital care and if the DPHN office cannot be contacted within a timely timeframe, arrangements will be made for the Parent(s)/legal Guardian(s) and the baby to return to the hospital for sample taking.

2.7.26 Public Health Nursing Repeat NNBS

2.7.26.1 The DPHN/Designated Officer will ensure the request for the repeat NBSS is forwarded to the RPHN/RM/RGN on the day of receipt of request for repeat NBSS.

2.7.26.2 The RPHN/RM/RGN will contact the Parent(s)/legal Guardian(s) to arrange for sample taking either at a home visit or at a clinic visit within 24hours.

2.7.26.3 The repeat NBSS will be completed on the day of receipt of notification or the following day (within 24hours).

Stage Three: Procedure for Repeat Sampling

2.7.27 Repeat NBS Sampling

2.7.27.1 The sample taker will advise the Parent(s)/legal Guardian(s) about the reason for the repeat.

2.7.27.2 The sample taker must:

- explain to the Parent(s)/legal Guardian(s) why a repeat sample has been requested.
- assure the Parent(s)/legal Guardian(s) that if the repeat sample should prove positive, that they will be contacted immediately by a liaison nurse /Clinical Nurse Specialist from the relevant maternity hospital/unit programme.
- be aware that the Parent(s)/legal Guardian(s) can decide to opt out of the repeat sample being taken. Section 2.7.13 covers this in detail.

2.7.27.3 The sample will be taken as per Stage Two –Section 2.7.15-2.7.19.

2.7.27.4 The sample taker must clearly indicate on the NBSC that the sample is a Repeat Sample by ticking the box 'Yes' and if the repeat is requested for a specific condition as per the eReport™, then the sample taker should specify in the comments box on the NBSC what the repeat is for.

2.7.27.5 The sample should then be sent immediately to the NNBSL either by registered post or by courier or fast track services as appropriate.

2.7.27.6 The sample taker informs the DPHN and/or NNBSL where relevant that the sample is taken and gives details of how the sample is sent to the laboratory.

2.7.27.7 The sample taker will keep a record of the NBST in the baby section of the maternal chart or the neo-natal baby chart for hospital services and/or the Child Health Record for community services.

Stage Four: NBSP Repeat Sample Results

2.7.28 NNBSL notify Maternity Hospital/Unit/Community of repeat sample results

2.7.28.1 The NNBSL informs the DOMN/ Maternity Hospital/Unit and the DPHN/CHO of the repeat sample results (individual report) of the screening via eReports™.

2.7.28.2 The NNBSL follows up on all children where a result is not normal and/or requires clarification and follow up.

2.7.28.3 The NNBSL liaises with the designated liaison nurse in the relevant maternity hospital/unit who refers the baby and their Parent(s)/legal Guardian(s) to the appropriate care pathway if required.

2.7.28.4 The Medical Team/Clinical Nurse Specialist for the appropriate care pathway makes contact with the family and arranges a hospital /outpatient visit for babies with a suspected positive result (See section 2.7.23).

Stage Five: Checking NNBSP Repeat Sample Results

2.7.29 Checking NBSP register – sample results

2.7.29.1 The designated officer for the DOMN/Maternity Hospital/Unit and/or CHO DPHN for the designated LHO continues to follow up on repeat sample results until screening is closed.

3.0 GOVERNANCE AND APPROVAL

3.1 Outline Formal Governance Arrangements

3.1.1 The National Newborn Bloodspot Screening Programme Governance Group will oversee the operation of the NNBSP (Appendix XXVI and XXVII). The aim of the NNBSP Governance Group is to oversee the development and implementation of an effective and efficient quality assurance framework for the NNBSP to ensure that all babies are offered newborn bloodspot screening in accordance with agreed protocols and standard operating procedures. This will be achieved by:

- Monitoring, auditing and evaluating the NNBSP using agreed key performance indicators and other quality assurance measures to facilitate improvements in the quality of the screening process and outcomes for parents/legal guardians and their babies
- Supporting the development of information and training resources for health professionals
- Engaging with parent/legal guardians, through representation on the NNBSP Governance Group, to develop information resources relevant to parents
- Providing multidisciplinary advice to CHO and Hospital Groups regarding technical and operational aspects of the NNBSP
- Provide advice to CHO, Hospital Groups and the Department of Health regarding strategic direction, policy and quality standards, funding objectives, legislation reviews and programme documentation such as referral guidelines, manuals for practitioners, guidelines for storage, retention and use of residual bloodspots etc.
- Following clear, agreed processes for assessing the inclusion of additional conditions into the NNBSP and resultant funding required to progress
- Advising the Department of Health on recommendations for expansion of the NNBSP
- Reporting to the Assistant National Director of Health and Wellbeing – Public Health and Child Health in relation to progress of the programme. The responsibility for this will lie with the Chairperson

3.1.2 The National Governance Group for Child Health Screening and Surveillance Programmes, chaired by the Assistant National Director in the Health and Wellbeing Division have a national governance and oversight role and the chair of the NNBSP Governance Group will be a member of this governance group (Appendix XXVIII). Any governance issues that arise with regard to the NNBSP can be brought to the attention of this group.

3.2 List method for assessing the PPPG in meeting the Standards outlined in the HSE National Framework for developing PPPGs.

This procedure was drafted by a subgroup of the NNBSP Governance Group using the HSE National Framework for developing PPPGs. It was initially drafted on the old template and is now been redrafted to be compliant with the new framework from developing PPPGs.

The membership of the NNBSP Governance Group consists of staff members that have extensive experience of the NNBSP from the Maternity/PHN services in addition to staff from the NNBSL.

This PPPG was reviewed by key personnel within the Primary Care Division, Health and

Wellbeing Division, Public Health Nursing, Maternity Hospitals and the NNBSL.

3.3 Attach any copyright/permission sought

Not applicable

3.4 Insert approved PPPG Checklist

Standards for developing Clinical PPPG	Checklist
Stage 1 Initiation	
The decision making approach relating to the type of PPPG guidance required (policy, procedure, protocol, guideline), coverage of the PPPG (national, regional, local) and applicable settings are described.	✔
Synergies/co-operations are maximised across departments/organisations (Hospitals/Hospital Groups/Community Healthcare Organisations (CHO)/National Ambulance Service (NAS)), to avoid duplication and to optimise value for money and use of staff time and expertise.	✔
The scope of the PPPG is clearly described, specifying what is included and what lies outside the scope of the PPPG.	✔
The target users and the population/patient group to whom the PPPG is meant to apply are specifically described.	✔
The views and preferences of the target population have been sought and taken into consideration (as required).	**Not Required**
The overall objective(s) of the PPPGs are specifically described.	✔
The potential for improved health is described (e.g. clinical effectiveness, patient safety, quality improvement, health outcomes, quality of life, quality of care).	✔
Stakeholder identification and involvement: The PPPG Development Group includes individuals from all relevant stakeholders, staff and professional groups.	✔
Conflict of interest statements from all members of the PPPG Development Group are documented, with a description of mitigating actions if relevant.	✔
The PPPG is informed by the identified needs and priorities of service users and stakeholders.	✔
There is service user/lay representation on PPPG Development Group (as required).	N/A
Information and support is available for staff on the development of evidence-based clinical practice guidance.	✔
Stage 2 Development	**Checklist**
The clinical question(s) covered by the PPPG are specifically described.	✔
Systematic methods used to search for evidence are documented (for PPPGs which are adapted/adopted from international guidance, their methodology is appraised and	Not Required – Programme

documented).	already in operation
Critical appraisal/analysis of evidence using validated tools is documented (the strengths, limitations and methodological quality of the body of evidence are clearly described).	Not Required – Programme already in operation
The health benefits, side effects and risks have been considered and documented in formulating the PPPG.	Not Required – Programme already in operation
There is an explicit link between the PPPG and the supporting evidence.	✔
PPPG guidance/recommendations are specific and unambiguous.	✔
The potential resource implications of developing and implementing the PPPG are identified e.g. equipment, education/training, staff time and research.	Not Required – Programme already in operation
There is collaboration across all stakeholders in the planning and implementation phases to optimise patient flow and integrated care.	Not Required – Programme already in operation
Budget impact is documented (resources required).	Not Required – Programme already in operation
Education and training is provided for staff on the development and implementation of evidence-based clinical practice guidance (as appropriate).	Not Required
Three additional standards are applicable for a small number of more complex PPPGs: Cost effectiveness analysis is documented. A systematic literature review has been undertaken. Health Technology Assessment (HTA) has been undertaken.	Not Required
Stage 3 Governance and Approval	**Checklist**
Formal governance arrangements for PPPGs at local, regional and national level are established and documented.	Previous draft SOP was approved in 2016 – no change
The PPPG has been reviewed by independent experts prior to publication (as required).	Not Required

Copyright and permissions are sought and documented.	Not Required
Stage 4 Communication and Dissemination	**Checklist**
A communication plan is developed to ensure effective communication and collaboration with all stakeholders throughout all stages.	✔
Plan and procedure for dissemination of the PPPG is described.	✔
The PPPG is easily accessible by all users e.g. PPPG repository.	✔
Stage 5 Implementation	**Checklist**
Written implementation plan is provided with timelines, identification of responsible persons/units and integration into service planning process.	Already in place
Barriers and facilitators for implementation are identified, and aligned with implementation levers.	Not Applicable
Education and training is provided for staff on the development and implementation of evidence-based PPPG (as required).	Not Required
There is collaboration across all stakeholders in the planning and implementation phases to optimise patient flow and integrated care.	Already in place
Stage 6 Monitoring, Audit, Evaluation	**Checklist**
Process for monitoring and continuous improvement is documented.	Already in place
Audit criteria and audit process/plan are specified.	Already in place
Process for evaluation of implementation and (clinical) effectiveness is specified.	Not required
Stage 7 Revision/Update	**Checklist**
Documented process for revisions/updating and review, including timeframe is provided.	✔
Documented process for version control is provided.	✔

4.0 COMMUNICATION AND DISSEMINATION

4.1 Describe communication and dissemination plans

This is the first version of this procedure developed by the NNBSP Governance Group.

4.1.1 The DOMN and DPHN will ensure that:

- Nursing and Midwifery staff members will be informed of the approval of this procedure by personal memo.
- All relevant administrative and clerical staff members will be informed of the approval of this procedure by personal memo.
- Nursing and Midwifery staff will be offered an educational update on the NNBSP when applicable from the NNBSL and the NNBSP Governance Group.
- This version of the procedure is placed in the "Policy, Procedure, Protocols and Guideline" folder in the Maternity Hospital/Unit Ward and Health Centre and is available to all staff members, from the date of implementation. It is the responsibility of Line Managers to ensure that staff members working under their remit have read and signed the policy in each clinical area.
- This version of the document will be made available in an electronic format.

4.1.2 The NNBSP Governance Group will ensure:

- This document is available on the newborn bloodspot screening website.
- The roll out of the HSE LanD online training module on Newborn Bloodspot Screening

5.0 IMPLEMENTATION

5.1 Describe implementation plan listing actions, barriers and facilitators and timelines

The NNBSP is already in operation using the "*Practical Guide to Newborn Bloodspot Screening in Ireland*" as a basis for its operation. Therefore there will be no requirement for an implementation plan. This Procedure will be disseminated as per Communication and Dissemination plan (Section 4)

5.2 Describe education/training plans required to implement the PPPG

The NNBSP is already in operation using the "*Practical Guide to Newborn Bloodspot Screening in Ireland*" as a basis for its operation. The HSE LanD online training module has been updated and will be offered to relevant staff in conjunction with the communication and dissemination of this procedure. It is envisaged that there will be education/study days organised also.

5.3 Identify lead person(s) responsible for the implementation of the PPPG.

The Assistant National Director Health and Wellbeing Division and the Clinical Director of the NNBSL will be jointly responsible for the overall operation of the NNBSP.

5.4 Outline specific roles and responsibilities.

5.4.1 The DOMN within Maternity Hospitals/Units is responsible for ensuring that all staff members working in the Maternity Hospital/Unit, who are responsible for requesting NBS sampling from the CHO, Public Health Nursing services/LHO, have read understand and adhere to this document and have signed the Signature Sheet to illustrate this (Appendix I). A record is kept of all staff members who have signed the Signature Sheet.

5.4.2 The Consultant Neonatologist and Consultant Paediatrician are responsible for ensuring that their staff members are aware of this procedure.

5.4.3 The DPHN within CHOs/LHOs is responsible for ensuring that all staff members working in the Public Health Nursing Service have read, understand and adhere to this procedure and have signed the Signature Sheet to illustrate this (Appendix 1). A record is kept of all staff members who have signed the Signature Sheet.

5.4.4 The DPHN liaises within the CHOs/LHOs and/or the Network Managers to ensure that all relevant administration/clerical staff members supporting the NNBSP have read, understood and adhere to this procedure and have signed the Signature Sheet to illustrate this (Appendix 1). A record is kept of all staff members who have signed the Signature Sheet.

5.4.5 The Public Health Doctors, Community Medical Doctors and GPs are responsible for ensuring staff members working within their services where applicable are aware of this procedure.

5.4.6 Each individual Midwife/Nurse, SECM, private Midwifery Group and Nursery Nurse undertaking the NNBSP in accordance with this procedure is responsible for reading, understanding and adhering to the contents of this procedure.

5.4.7 The designated Midwifery Officer will ensure that all SECMs who are employed under the Memorandum of Understanding and Contractual Agreement (2014) have read, understood and adhere to this procedure and have signed the Signature Sheet to illustrate this (Appendix I). A record is kept of all staff members who have signed the Signature Sheet.

5.4.8 Midwifery and Nursing staff members working in the Public Health Nursing Service within CHOs/LHOs are responsible for ensuring that they work within their scope of practice (Scope of Nursing and Midwifery Practice Framework, 2015).

5.4.9 The **Health Service Executive** has ultimate responsibility for ensuring that all babies are offered screening in accordance with agreed protocols and procedures. Within the HSE this responsibility lies with the National Director of Health and Wellbeing. The operation of the NNBSP is the responsibility of Assistant National Director of Health and Wellbeing – Public Health and Child Health. The Director of the NNBSL is responsible for the day to day coordination of the NNBSP.

5.4.10 The **CHO Child Health Lead** has the authority and responsibility of ensuring that **all babies born and all newborns residing in their CHO** area are offered screening and that the structures for the timely checking and recording of the test results are in place. The CHO Child Health Lead is responsible for reporting newborn bloodspot screening uptake and coverage to the Chief Officer of the CHO and the Assistant National Director of Health and Wellbeing – Public Health and Child Health and the NNBSP Governance Group.

5.4.11 **Directors of Midwifery/Nursing of Maternity Hospitals/Units** are responsible for ensuring that all babies born in hospital are offered the newborn bloodspot screening test. If the test is not performed in the Maternity Hospital/Unit before discharge, hospital staff are responsible for ensuring that the baby is screened either by returning to the Maternity Hospital/Unit, or by informing the Public Health Nursing service and/or Hospital Community Midwifery Service or the SECM of the discharge and the requirement for newborn bloodspot screening.

5.4.12 **Directors of Public Health Nursing in the CHOs/LHOs** are responsible for ensuring that the newborn screening test is carried out in the local health area if required following notification from the Maternity Hospital/Unit and that all babies residing in their designated area have been offered the NNBSP. The Directors of Public Health Nursing are responsible for the timely recording of all babies' (hospital and community) newborn bloodspot screening test results in the newborn screening register including newborn bloodspot screening results for babies who require follow-up and further investigation.

5.4.13 **Self Employed Community Midwives (SECMs) and Private Midwifery Groups** are responsible for performing the NNBSP in accordance with agreed protocols and procedures and for dispatching the newborn bloodspot screening card to the NNBSL as soon as possible after collection in accordance with packaging transport regulations. The designated Midwifery Officer is responsible for monitoring the practice of SECMs in accordance with the Memorandum of Understanding and Contractual Agreement (2014).

5.4.14 **Parent(s)/Legal Guardian(s)** are ultimately responsible for their baby and their participation in the NNBSP.

- In the case of Parent(s)/Legal Guardian(s) opting out of the NNBSP and having been informed by practitioners of the potential consequences to their baby in so doing, the responsibility for the possible adverse consequences lies with the Parent(s)/Legal Guardian(s). Parent(s)/Legal Guardian(s) may change their mind in the future but it is their responsibility to bring this 'change of mind' to the attention of PHN and/or GP.
- In the case of babies born outside the jurisdiction, the Parent(s)/Legal Guardian(s) are responsible for ensuring that the baby is screened. If the Parent(s)/Legal Guardian(s) notify the RPHN of the birth on the return to this jurisdiction, the PHN is responsible for ensuring that the newborn bloodspot screening test is carried out.

6.0 MONITORING, AUDIT AND EVALUATION

6.1 Describe the plan and identify lead person(s) responsible for the following processes:

6.1.1 Monitoring

6.1.1.1 The NNBSP Governance Group will oversee a performance management framework that will:

- Enable health care professionals to ensure that the services they provide meet core standards for that service
- Develop and share information on good practice and to further improve quality of service as a consequence
- Provide accessible and clear information for the users of the service to enable them to make better informed decisions about their care.

6.1.1.2 In relation to the NNBSP, process standards and performance indicators have been developed in relation to the following:

- Timely sample collection
- Timely sample dispatch
- Completeness of coverage
- Enhanced tracking abilities
- Timely identification of babies for whom the laboratory has not received a decline notification or a blood sample
- Timely processing of positive screening samples

6.1.2 Audit

6.1.2.1 Midwifery/Nursing/Public Health Nursing management team will conduct an annual audit in line with the Quality Assurance Management Tools

6.1.3 Evaluation.

7.0 REVISION/UPDATE

7.1 Describe procedure for the update of the PPPG (including date for revision).

A formal review of this version will be carried out on a six monthly basis initially and will then be carried out on a three yearly basis unless there is a change required informed by legislation, best practice or any relevant EU Directives which may indicate a requirement to update this procedure sooner.

The responsibility for this review lies with the NNBSP Governance Group. Any learnings that arise from the ongoing monitoring and evaluation of the NNBSP that may impact on this procedure will be used to amend, update and change the original procedure version. If there are no amendments to the PPPG following the review process, the date and detail on the version tracking box on the front cover of the PPPG will be updated anyway.

7.2 Identify method for amending PPPG if new evidence emerges.

All healthcare professionals participating in the NNBSP will be aware, through continuous professional development activities, such as conferences, journal papers etc., of any new evidence regarding best practice that emerges in the area of newborn bloodspot screening. The health professionals will be obliged to inform the relevant Child Health Lead in their area of this change in evidence or best practice and the NNBSP Governance Group should be informed.

7.3 Complete version control update on PPPG Template cover sheet.

8.0 REFERENCES

HSE (2016) A Practical Guide to Newborn Bloodspot Screening in Ireland – 7th Edition. National Newborn Bloodspot Screening Laboratory, Children's University Hospital Temple Street

9.0 APPENDICES

Appendix I:

Signature Sheet

I have read, understand and agree to adhere to this Policy, Procedure, Protocol or Guideline:

Print Name	Signature	Area of Work	Date

Appendix II:

Membership of the PPPG Development Group (Template)

Please list all members of the development group (and title) involved in the development of the document.

Barbara Bolger National Specialist Primary Care Operations	Signature: _____ Date: _____
Grace O'Neill Regional Child Health Training/Development Officer/Immunisation Co-Ordinator	Signature: _____ Date: _____
Paul Marsden Project Manager Child Health Screening Programmes	Signature: _____ Date: _____
Loretta O'Grady Chief Medical Scientist, National Newborn Bloodspot Screening Laboratory	Signature: _____ Date: _____
Dr. Ingrid Borovickova Clinical Director, National Newborn Bloodspot Screening Laboratory	Signature: _____ Date: _____
Dr. Abigail Collins Consultant in Public Health Medicine	Signature: _____ Date: _____
Mary Finn-Gilbride Director of Public Health Nursing Wexford	Signature: _____ Date: _____
Colette McSweeney Assistant Director of Public Health Nursing	Signature: _____ Date: _____
Chairperson: Dr. Phil Jennings Director of Public Health National Lead National Healthy Childhood Programme Chairperson National Newborn Bloodspot Screening programme Governance Group	Signature: _____ Date: _____

Appendix III: Conflict of Interest Declaration Form

CONFLICT OF INTEREST DECLARATION

This must be completed by each member of the PPPG Development Group as applicable

Title of PPPG being considered:

Standard Operating Procedure for Maternity Hospitals/Units & Primary Care Services Delivering the National Newborn Bloodspot Screening Programme (NNBSP)

Please circle the statement that relates to you

1. I declare that I DO NOT have any conflicts of interest.

2. I declare that I DO have a conflict of interest.

Details of conflict (Please refer to specific PPPG)

_____

_____

_____

_____

(Append additional pages to this statement if required)

Signature

Printed name

Registration number (if applicable)

Date

The information provided will be processed in accordance with data protection principles as set out in the Data Protection Act. Data will be processed only to ensure that committee members act in the best interests of the committee. The information provided will not be used for any other purpose.

A person who is covered by this PPPG is required to furnish a statement, in writing, of:

(i) The interests of the person, and

(ii) The interests, of which the person has actual knowledge, of his or her spouse or civil partner or a child of the person or of his or her spouse which could materially influence the person in, or in relation to, the performance of the person's official functions by reason of the fact that such performance could so affect those interests as to confer on, or withhold from, the person, or the spouse or civil partner or child, a substantial benefit.

Appendix IV:

Membership of the Approval Governance Group

Please list all members of the relevant approval governance group (and title) who have final approval of the PPPG document.

Note: This PPPG was approved in August 2016 by Primary Care and Health and Wellbeing Operations as per title page. There was a delay in implementation the PPPG due to a number of clarifications required due to changing organisational structures and personnel. There is a short revision date on this PPPG and an appropriate Approval Governance Group will be convened from with the new structures at that time.

Type Name here	Signature: _____
Type Title here	Date: _____
Type Name here	Signature: _____
Type Title here	Date: _____
Type Name here	Signature: _____
Type Title here	Date: _____
Type Name here	Signature: _____
Type Title here	Date: _____
Chairperson:	
Type Name here	Signature: _____
Type Title here	Date: _____

Appendix V: Screening Conditions - Definitions

Taken from "*A Practical Guide to Newborn Bloodspot Screening in Ireland, 7th Edition (2018, pages 11-18*)"

All conditions which form part of the National Newborn Bloodspot Screening Programme, fulfil, in part or in full, the criteria which have been set out internationally for newborn screening. These include:

- the conditions screened are treatable;
- there is a test available which is easily applied to large population groups;
- there are few false positive and false negative results i.e. the test is reliable;
- the incidence of the conditions in the community is sufficiently high to warrant screening;
- the cost of screening makes the process cost-effective

For all of the conditions, early diagnosis and treatment significantly improves the clinical outcome. Some of the conditions, for example, Classical Galactosaemia and Maple Syrup Urine Disease benefit from the earliest detection possible. Unfortunately, as with all screening programmes, not all individuals with a condition will be detected. This is particularly true for Homocystinuria where about one in five babies may not be detected by screening and for Cystic Fibrosis where milder variants of the condition may not be detected.

N.B.: No screening test is 100% reliable

Conditions included in the Irish NNBSP

Condition	Date Started	Irish Incidence	Worldwide Incidence
Phenylketonuria (PKU)	1966	1: 4,500	1: 12,000
Homocystinuria (HCU)	1971	1: 69,400	1: 120,000
Classical Galactosaemia (CGal)	1972	1: 16,200	1: 45,000
Maple Syrup Urine Disease (MSUD)	1972	1: 155,200	1: 225,000
Congenital Hypothyroidism (CHT)	1979	1: 2,300	1: 3,500
Cystic Fibrosis (CF)	2011	1: 2,300	1: 3,500
Medium Chain Acyl-CoA Dehydrogenase Deficiency	2018	1:66,000	1:14,600
Glutaric Aciduria type 1	2018	1:54,000	1:100,000

Mode of Inheritance

The majority of the conditions involve a defect in a metabolic process or pathway and are inherited as 'autosomal' conditions, not being dependent on the gender or sex of the individual.

Each step in a metabolic pathway is governed by an enzyme, a protein produced by a set (two) of genes on a pair of chromosomes. Each parent transfers to their off-spring one set of genes so that the off-spring has a set from each parent. If one of these genes is defective (mutated), the metabolic pathway continues, albeit at a reduced rate. These individuals, known as carriers, do not have symptoms of the condition but carry the defective gene. If both parents are carriers, their offspring have a one in four chance of having the condition. For autosomal recessively inherited conditions, both parents must be carriers of a defective gene and each of their off-spring has a one in four chance of having the condition and a one in two chance of being a carrier.

The clinical presentation and severity of the condition may vary between unrelated families. Different mutations may affect the same metabolic process differently; some individuals may present with a severe form of the condition and others with a very mild form.

Congenital hypothyroidism is slightly different in that for 90% of cases the condition just occurs, for reasons which are not fully understood; it is twice as common in girls as in boys. However, for one in 10 babies (10%) the condition is inherited as an autosomal recessive condition, as described above.

Conditions included in the Screening Programme

Phenylketonuria (PKU)

Phenylketonuria is an autosomal recessive condition involving the breakdown of the amino acid phenylalanine. Approximately one in every 4,500 babies born in Ireland have PKU or a milder form called hyperphenylalaninaemia. When diagnosed within the newborn period and started on treatment, these babies grow up healthy and well. However, without treatment this condition may cause intellectual or physical disability.

In the majority of cases, the condition is caused by a lack of the enzyme phenylalanine hydroxylase, which normally converts phenylalanine, one of the building blocks in protein, into tyrosine. In the absence on the enzyme, phenylalanine accumulates and high levels have a direct toxic effect on the brain.

Early treatment is very beneficial; it aims at giving a reduced intake of phenylalanine but a normal intake of all the other amino acids. This diet has to be continued for life. The screening test depends on detecting a high level of phenylalanine in the blood. If the test is carried out before about 72 hours after birth, there is a possibility that the level of phenylalanine in blood may not be sufficiently elevated for the condition to be detected.

The treatment for PKU has been one of the major successes in medicine since it was first introduced in the early 1950s. There is substantial evidence to show that the earlier treatment is started and the better the biochemical control throughout life, the better the outcome.

Maple Syrup Urine Disease (MSUD)

Maple Syrup Urine Disease is a life threatening condition if it is not detected and treated early. It too is an autosomal recessive condition caused by a defect in the metabolism of three amino acids, known as the branched chain amino acids because of their similar biochemical structure. Approximately one in every 155, 200 babies born in Ireland may have this condition or about one baby born every two to three years. The disorder is so called because the urine may have an odour similar to that of maple syrup.

Screening was originally justified on the basis that chronic handicap and even premature death had occurred in a number of families where this condition had gone undetected. The branched chain amino acids accumulate in blood following the establishment of feeding during the first few days of life, and may cause brain damage.

A diet similar to that for PKU but with low levels of the branched-chain amino acids is started as soon as the diagnosis is made. Normal brain development and good health result from early treatment; life-long adherence to the diet is essential. Urgent medical intervention may be required during illness, which may be precipitated by infection or stress.

Homocystinuria (HCU)

Homocystinuria results from the accumulation in blood of the amino acid methionine and one of its metabolic products homocysteine. Homocysteine accumulates due to a deficiency of the enzyme cystathionine β-synthase. Homocysteine is toxic to the lining of blood vessels and predisposes the individual to thrombus formation, blood clots and a number of other complications including osteoporosis (thinning of the bones) and dislocation of the lens of the eye. Again the treatment is similar to that for PKU. For those individuals who adhere to the diet, the risk of developing any of the complications is greatly reduced. Approximately one in every 69,400 babies born in Ireland may have the condition or one baby every year.

The screening programme detects high blood levels of methionine. This is one of the more difficult conditions to detect, as the blood methionine level may not be raised initially. The methionine concentration is low in many baby foods, particularly in breast milk. The screening programme may not detect approximately one in every five babies born with this condition. There are a variety of reasons why this may occur. These include:

- Breast fed babies as there may be an inadequate intake of methionine in the feed to enable the blood methionine level to rise above the level for diagnosis.
- A milder vitamin B6 responsive form of the condition. These patients usually have milder symptoms and disease progression is slower and they are unlikely to be detected by newborn screening.

Consequently, if protein intake is deemed to be suboptimal, a further sample should be taken on or about day 10 of life for Homocystinuria screening. All babies or children who present clinically in later life with signs and symptoms suggestive of Homocystinuria, such as dislocation of lenses, osteoporosis or inappropriate tall stature should have the disorder excluded formally by measuring plasma levels of methionine and total homocysteine using a standard amino acid analyser.

Classical Galactosaemia (CGal)

Classical Galactosaemia is an autosomal recessive condition caused by the deficiency of the enzyme galactose-1-phosphate uridyl transferase. This enzyme is important for the breakdown of galactose, one of the two sugars that make up lactose in human and cow's milk. Approximately 1 in every 16,200 babies born in Ireland each year may have this condition. However, it is particularly common among babies born to Irish Traveller parents in whom the incidence is approximately 1 in 450 births. Consequently, in the non-traveller Irish community the incidence is about one in every 36,000 births.

If not detected and treated during infancy, the disorder may cause damage to the liver or there may be an increased risk of infection, which may be life threatening. As a result of the condition, galactose and its metabolite galactose-1-phosphate accumulate in blood. Galactose-1-phosphate is extremely toxic. The baby may present with jaundice and there may be a bleeding disorder with a tendency to bleed spontaneously. The affected baby may also develop an *E coli* infection of the blood, septicaemia or present with cataracts in their eyes. Early detection and treatment with a lactose or galactose-free diet will prevent the early clinical complications of the disorder; some of the longer term complications, such a dyspraxia, ataxia or reduced fertility in women, may still occur in older children and adults despite dietary treatment.

Because the condition is more common in babies born to Irish Traveller parents and to siblings of known cases, a special screening test, the Beutler test, is offered to all these babies at birth (preferably Day 1 of life). Parents/legal guardians are advised to keep baby on a galactose free feed (Soya-based) until the result of the Beutler test is available. This protects the baby should he/she have the condition. For those mothers wishing to breast feed, they should discuss this with their midwife as they can express their milk until the result of the Beutler test is available.

Clinicians should never depend upon the general population screening for the diagnosis of Classical Galactosaemia, but should query this condition in any baby who presents early with jaundice and other symptoms suggestive of Galactosaemia e.g. vomiting, floppiness, hypoglycaemia, conjugated hyperbilirubinaemia or abnormal clotting of unknown cause.

Cystic Fibrosis (CF)

Ireland has one of the highest incidences of CF in the World with approximately one in every 2,300 babies being affected. CF is also an autosomal recessive condition with both parents carrying an abnormal Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene.

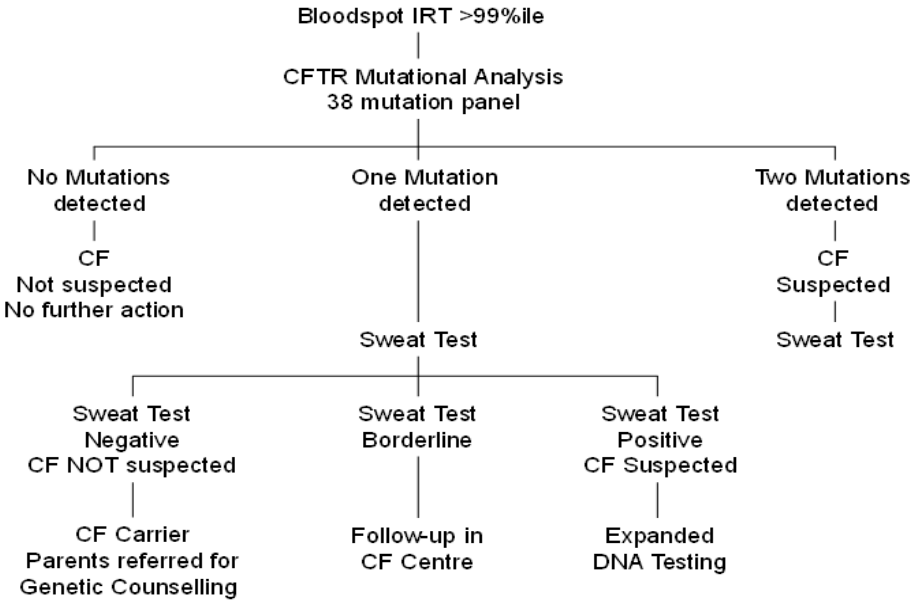
As a consequence of the condition, thick mucus secretions are produced by a number of organs including the lungs and pancreas; it is this thick mucus secretion which causes the problems. The thick secretions in the lungs may become infected, ultimately causing damage to the lungs. If the pancreas is involved this may cause diabetes mellitus, digestive problems and malabsorption of important vitamins. Consequently babies with CF may present with failure to thrive and frequent chest infections.

Newborn blood spot screening means that babies with CF are identified earlier; they can be treated with a high energy diet to improve weight gain and medicines and physiotherapy to improve lung function. Although a child with CF may still become ill, early treatment does improve their quality of life, significantly reducing the time that they have to spend in hospital; they can live healthier and longer lives. Specific treatment is now available for a significant number of persons with CF depending on the specific mutation affecting the CFTR gene.

The screening programme measures the blood level of immunoreactive trypsinogen (IRT). IRT is normally excreted by the pancreas into the intestinal tract, but in individuals with CF, it is regurgitated back into the blood due to the thick mucus secretions which block the pancreatic ducts. Levels of IRT may remain high in blood for about the first six weeks of life. If the blood IRT level is high the sample will be referred for CFTR mutational analysis. This DNA test screens for the presence of 38 possible mutations on the original bloodspot collected.

- If two CF mutations on the newborn screening sample are identified, then the baby probably has CF and this will be confirmed by a sweat test which will be organised and carried out by the babies local CF centre without delay.
- If one mutation is identified then a sweat test will be performed to determine whether the baby is a carrier of CF or has the condition. If the sweat test is positive, further DNA analysis will be undertaken to identify the second mutation.

Figure 1: Algorithm for Newborn Screening for Cystic Fibrosis



The 'sweat test' used to confirm or out-rule CF, measures the chloride concentration in sweat, and is usually performed before the fourth week of life in one of six designated HSE paediatric CF centres, based on the baby's address. The sweat test is considered the 'gold' standard for confirming the diagnosis of CF. As with some of the other conditions included in the screening programme, not all babies with CF will be detected by the newborn bloodspot screening programme. Milder variants of the condition may not be detected; some of these individuals may have a very benign clinical course which may not require treatment.

Note

- Newborn dried blood spot IRT screen for CF is not suitable for babies/children over six weeks of age.
- Babies with meconium ileus (MI) should be strongly suspected of having CF and followed up accordingly, including CF mutations at birth. These babies may have a normal CF screen (normal IRT). MI must be noted on the screening card.
- The Irish CF mutational genetic panel screens for 38 mutations chosen to reflect the Irish population, therefore babies of non-Irish ethnic origin are at increased risk of non-detection within the Irish programme, particularly if parents are consanguineous.

Congenital Hypothyroidism (CHT)

Unlike the other conditions, CHT is a congenital rather than an inherited condition, in the majority of cases. This is an endocrine condition, which results from failure of the thyroid gland to produce the hormone thyroxine. There are a number of different forms of the condition. Some babies may have a very small thyroid gland or no gland at all while others may not be able to make thyroxine. It is important to identify the cause; this can be done by performing a thyroid scan soon after the diagnosis has been made and usually before treatment has been started. Approximately 1 in every 2,300 babies born in Ireland may have the condition; early detection allows for early treatment and the prevention of symptoms.

The diagnosis is made by measuring blood thyroid stimulating hormone (TSH), high levels of which are

suggestive of the condition. However, TSH rises in blood immediately after birth and then falls to normal by about the second day of life. This is one of the reasons why the heel-prick sample should not be taken before 72 hours after birth, otherwise a false-positive result for congenital hypothyroidism may occur.

The majority of babies with congenital hypothyroidism require thyroid hormone replacement. Some babies will be reviewed between two and three years of age at which time a small number may be able to discontinue treatment under medical supervision. Otherwise treatment is for life and the dose of thyroxine adjusted as the baby grows.

Compared to some of the other conditions, the frequency of false positive results for CHT is relatively high. Consequently the number of requests for repeat blood samples is also high. Possible reasons include:

- A transiently raised TSH concentration, which returns to normal in time. These babies may require a number of repeat samples to be collected.
- Hypothyroidism is more common in babies and children with Down Syndrome, as a result a disproportionate number of repeat samples may be requested from these babies as they may have a transiently elevated plasma TSH level during the newborn period before developing hypothyroidism later.
- Babies, who have had surgery before having the screening sample taken, may have a transiently elevated plasma TSH level. This may occur as some antiseptic skin preparations contain iodine which may be absorbed through the skin and cause transient hypothyroidism. This occurs more commonly in premature babies.

Medium Chain Acyl CoA Dehydrogenase deficiency (MCADD)

MCADD is an autosomal recessive inherited defect of fatty acid oxidation due to deficiency of the enzyme medium-chain acyl-CoA dehydrogenase. This enzyme is required for the metabolism of medium-chain fatty acids and is necessary to enable the body to use its own fat reserves to produce energy in periods of fasting or stress.

Symptoms are not apparent at birth and about one third of cases of MCADD remain asymptomatic throughout life, however, symptoms can develop very quickly in affected infants who are not feeding well. Complications typically arise during periods of stress caused by an illness, fasting or vomiting, when the infant needs to break down fat quickly.

Episodes of metabolic decompensation can be prevented through avoidance of fasting, and monitoring of the infant to determine 'safe' time periods between meals and following a strict feeding schedule. MCADD mainly presents before the age of two years with a mean age of thirteen months, although neonatal presentations have also been reported.

Hypoglycaemia and a decompensated state develop which can result in serious life threatening symptoms including seizures and brain damage. When diagnosed within the newborn period and started on treatment, these babies grow up healthy and well. However, without treatment this condition may cause intellectual or physical disability or even death

Glutaric aciduria type 1 (GA1)

Glutaric aciduria type 1 (GA1) is an autosomal recessive inherited condition caused by a deficiency of the enzyme glutaryl-CoA dehydrogenase (GCDH). With this condition the body is unable to break down certain proteins properly. It is an organic acid condition as it can lead to harmful amounts of organic acids and toxins in the body. If left untreated it can cause brain defects or even death, however if the condition is detected early in life and proper treatment begins children with GA1 can lead healthy lives.

The enzyme GCDH is involved in the decarboxylation of glutaryl-CoA, which is an intermediate in the breakdown of the amino acids lysine, hydroxylysine and tryptophan. Defective catabolism causes the toxic accumulation of glutaric acid, 3-hydroxyglutaric acid, glutaconic acid and glutaryl carnitine.

Over 150 disease causing mutations have been identified; of these the R402W mutation is the most prevalent among Caucasians. Most mutations, including the R402W mutation, are associated with undetectable GCDH activity and excretion of high amounts of glutaric acid. However, mutations that lead to varying levels of residual GCDH activity and low excretion of glutaric acid have also been reported. Consequently, patients with GA1 can be divided into two biochemically defined subgroups low or high excretors, based on the levels of glutaric acid present in the urine.

About 70% of patients (including both low and high excretors) have an encephalopathic crisis, which is most common at around nine months, with 90% by age two years. These are usually precipitated about 1–3 days after onset of a non-specific intercurrent illness, gastrointestinal infection or pneumonia and lead to dystonia and dyskinesia as permanent sequelae but with relative preservation of the intellect.

Appendix VI: DOMN/Maternity Hospital/Unit Unique Perinatal Identifier (UPI)

National Newborn Bloodspot Screening Programme - Unique Perinatal Identifier

What is the Unique Perinatal Identifier (UPI)

From 1st July 2011 each baby born in Ireland will be issued with an individual (unique) identifying number to ensure that Newborn Bloodspot Screening samples can be traced throughout the screening process.

Why is the UPI being introduced now?

This date has been chosen to coincide with the introduction of Newborn Screening for Cystic Fibrosis (CF) on 1st July 2011. CF screening will be added to the conditions for which screening already takes place through the 'heel-prick' method and will be carried out on the same bloodspot sample.

How will the UPI be issued?

The UPI will be formed by the 3 digit hospital HIPE code (see list of maternity hospitals/units below) of the birth hospital followed by the Healthcare Record Number (HCRN) of the baby.

Will a baby who is not born in an Irish Maternity Hospital/Unit receive a UPI?

Babies born either at home or in a maternity hospital/unit outside Ireland will be issued a UPI by the Director of Public Health Nursing in the area in which their birth is registered following notification of birth.

How will the UPU work?

The number will be used on all information relating to the National Newborn Bloodspot Screening Programme (NNBSP) including the following:

- On all Newborn Screening Cards sent to the National Newborn Screening Laboratory in the Children's University Hospital Temple Street including repeat samples
- On maternity hospital/unit Newborn Bloodspot Screening Registers
- On requests from maternity hospital/unit to Public Health Nursing Services to undertake Newborn Bloodspot Screening
- On requests from maternity hospitals/unit to a Children's hospital/unit to undertake Newborn Bloodspot Screening on babies transferred prior to completion of the NNBSP
- On LHO/ISA community child health service Newborn Bloodspot Screening Registers
- On NNBSL reports
- On all notifications of births from maternity hospitals/units to LHO/ISA community child health services (This may not be possible in all areas until the new Maternal and Newborn Clinical Management System is in place).

Will there be a space to put the UPI on the NBS card?

The Newborn Bloodspot Screening Cards have been modified to include a space for UPI.

What happens with maternity hospitals/unit where baby does not have its own healthcare record number?

For the small number of hospitals/units where this is the case, support has been provided through the Newborn Screening for Cystic Fibrosis Implementation process over recent months to assist in addressing the changes required prior to 1st July 2011.

HIPE Code	Maternity Hospital / Unit
201	Midland Regional Hospital Portlaoise
202	Midland Regional Hospital Mullingar
301	University Hospital Limerick
402	Cavan General Hospital
500	Letterkenny General Hospital
501	Sligo University Hospital
600	University Hospital Waterford
601	St Luke's General Hospital Kilkenny
605	Wexford General Hospital
607	South Tipperary General Hospital
724	Cork University Hospital
726	University Hospital Kerry
800	Galway University Hospitals
802	Mayo University Hospital
919	Portiuncula Hospital Ballinasloe
922	Our Lady of Lourdes Hospital Drogheda
930	Coombe Women and Infants University Hospital
931	National Maternity Hospital Holles Street
932	Rotunda Hospital Dublin

Appendix VII: Information for Parents and Guardians

In the first week after your baby is born, you will be offered newborn bloodspot screening for your baby. This is often called the 'heel prick'.

Newborn bloodspot screening is an essential part of newborn care. It helps identify babies who may be at high risk of having a rare but serious condition. Most babies who are screened will not have any of these conditions. But for the small number of babies who do, the benefits of screening are enormous.

What conditions are included in newborn bloodspot screening?

In Ireland, all babies are now screened for:

- Cystic fibrosis
- Congenital hypothyroidism
- Phenylketonuria
- Maple syrup urine disease
- Homocystinuria
- Classical galactosaemia
- Glutaric aciduria type 1
- Medium chain acyl CoA dehydrogenase deficiency

You can read more about these rare conditions on www.newbornscreening.ie. You can also discuss these with your midwife or public health nurse.

Why would my baby have one of these conditions?

Most of these conditions are inherited. This means the baby gets the genes that cause the condition from their parents. This also means there is a risk that other babies born to these parents may have the same condition.

Why should I have my baby screened?

Each year, newborn bloodspot screening identifies about 110 babies with one of these rare but serious conditions. The health of these babies can then be managed before they develop severe symptoms. Unmanaged, these conditions can cause a serious risk to health or life.

Some parents worry that their baby will be uncomfortable during the heel prick. But the long-term benefit of screening is much greater than the small discomfort a baby feels when the blood sample is taken.

When is the screening done?

Newborn bloodspot screening is usually done between three and five days after your baby is born.

How is the screening done?

The midwife or public health nurse will prick your baby's heel with a sterile needle to collect drops of blood onto a special card. They will then hold your baby's ankle to ensure the blood goes onto the card. This may take a couple of minutes. Your baby may feel uncomfortable and may cry. You can help by making sure your baby is warm and comfortable by cuddling and feeding them.

What happens after the heel prick is done?

When the sample is collected, the card is sent to the National Newborn Bloodspot Screening Laboratory at the Children's University Hospital, Temple Street, Dublin.

What happens to my baby's screening card after screening?

After screening, the results and the card are stored securely by Children's University Hospital, Temple Street, as part of your baby's health record. This is in line with Department of Health policy. The stored cards may be used for:

- checking your baby's results or for other tests that your doctor recommends, but only after you give your permission, and
- improving the screening programme and the health of babies and their families in Ireland.

Newborn bloodspot screening cards are sometimes used in research but never for commercial gain. When cards are used, babies are never identifiable.

Will my baby need to be screened more than once?

Sometimes a screening result is not clear or not enough blood was collected. If this happens, your midwife or public health nurse may need to contact you and ask to take a second blood sample from your baby's heel.

How will I hear about the results?

If the screening shows that your baby is not at high risk of having any of the conditions, you will not be contacted. If you would like a copy of the screening results, you can ask your public health nurse for them at your next visit.

If the screening shows your baby is at high risk of having one of these conditions, a nurse or doctor will contact you as soon as possible.

What happens if the screening says my baby is at risk?

If the screening shows your baby is at high risk, they will need to have more tests to confirm whether or not they do have the condition. They may need to stay in hospital for a short time while this is done.

How good is newborn bloodspot screening at finding babies at high risk of having one of these conditions?

Newborn bloodspot screening does not make a diagnosis. It shows only that a baby is '**at high risk**' of having one or more of the conditions screened for.

Sometimes screening results can suggest a baby is at high risk of having one of the conditions, but when more tests are done the baby actually does not have the condition. This is called a '**false positive**'. False positives can be very worrying for parents and families, but they are very rare.

Sometimes the screening result does not identify a possible health risk. This is called a '**false negative**'. A false negative means that the screening result does not show that a baby is at high risk, but the baby may actually have one of these conditions. False negatives are extremely rare. But if you have any concerns about your baby, discuss them with your family doctor (GP) or public health nurse.

I would like to have my baby screened. What should I do?

Your midwife or public health nurse will talk to you about screening and give you information. Please read the information carefully. If you have questions, please ask your midwife or public health nurse.

If you want to have your baby screened, sign the newborn bloodspot screening card you are given. Signing this card is how you confirm that the information about your baby is correct and how you agree (consent) to the screening.

What if I feel unsure about this screening?

If you are unsure about screening, please talk with your midwife or public health nurse. They will be able to discuss your concerns and explain more about the screening.

If you are still unsure, your midwife or public health nurse will offer you the opportunity to speak with a senior officer in the National Newborn Bloodspot Screening Laboratory in the Children's University Hospital, Temple Street.

If you decide not to get your baby screened, you will be asked to sign a form that says you understand the risks of not having your baby screened. If you change your mind, please talk to your public health nurse or family doctor (GP). They can arrange to have your baby screened.

Where can I get more information?

For more information on newborn bloodspot screening:

- visit the website www.newbornscreening.ie
- talk to your midwife or public health nurse

Plain English
Approved by NALA

Copies of the leaflet are available to order from www.healthpromotion.ie

Appendix VIII: National Maternity Hospitals/Units Discharge Checklist for Babies

DISCHARGE CHECKLIST: BABY

Baby healthcare record number _____

Gender: Boy ☐ Girl ☐

Is baby going home to address on label? Yes ☐ No ☐

Address baby discharged to (if different)

_____

_____ **Contact Numbers** _____

GP details _____

Please affix woman's ID label to all pages of checklist.
Label should contain the following information:
- Name
- Address
- Date of Birth
- Healthcare Record Number

To be filled in as appropriate during inpatient stay and checked at time of discharge to ensure all items completed prior to discharge

			Initials	Date / Time (24 hour)
NATIONAL NEWBORN BLOODSPOT SCREENING PROGRAMME ** NBs = Newborn Bloodspot* Unique Perinatal Identifier _____ _____ _____ - _____ Date NBs* taken: _____ **If not taken**, date due: _____ *If NBs taken, ensure that Nurse copy of screening card is filed in Baby notes.* **Is a second / subsequent NBs required?** _____ **If so, when?** _____ **If NBs to be taken following discharge, identify plan for completion.** Please tick (✓) relevant. PHN at baby's home ☐ Hospital baby clinic ☐ Health centre ☐ Return to ward ☐ Has parent / guardian chosen to opt out and signed **opt out form**? Yes ☐ No ☐				
ADVICE GIVEN REGARDING:	Please tick (✓) where relevant	**Comment (if required)**		
Changing / Top and tail / Handling	☐			
Cord care	☐			
Eye care	☐			
Bathing	☐			
Prevention of SIDS	☐			
Signs of effective feeding	☐			
Plagiocephaly	☐			
Other *(Please specify)*	☐			
DISCHARGE INFORMATION Method of feeding on discharge: Partial breastfeeding ☐ Discharge weight _____ grams *(Check percentage weight loss and if feeding assessment required)*	Please tick (✓) where relevant Exclusive breastfeeding ☐ Artificial ☐			
Are all baby observations satisfactory? *Any concerns (e.g. weight, feeding problems, jaundice)*				
Is baby currently on medication / due to complete course? of medication? *E.g. oral Vitamin K, anti-virals. Give details.*				
Has baby had BCG? Yes ☐ No ☐ Has baby had neonatal hearing screening? Yes ☐ No ☐ Has Paediatric Discharge been completed? *(Check notes)* ☐				
BABY FOLLOW-UP GP ☐ Baby clinic ☐ Consultant clinic ☐ *(Specify)* _____ Orthopaedic ☐ Physiotherapy ☐ Other ☐ *(Specify)* _____	Please tick (✓) where relevant			
Identity bracelets removed ☐ **Security tag removed** ☐				
Signature of midwife completing discharge			**Date**	**Time (24 hour)**
Name in PRINTED BLOCK CAPITALS				

Triplicate form: 1st copy to mother for PHN, 2nd copy retained in mother's notes, 3rd copy filed in baby's notes.

NMC 9 Disch Chkl B v.1

Appendix IX: DPHN/CHO/LHO Unique Perinatal Identifier for NNBSP

Unique Perinatal Identifier (UPI) for non-Irish hospital births.

The first 3 digits of the UPI for non-Irish hospital births will be as follows:

L01	Dublin Area 1
L02	Dublin Area 2
L03	Dublin Area 3
L04	Dublin Area 4
L05	Dublin Area 5
L06	Dublin Area 6
L07	Dublin Area 7
L08	Dublin Area 8
L09	Wicklow
L10	Kildare
L11	Louth
L12	Meath
L13	Cavan
L14	Monaghan
L15	Longford/Westmeath
L16	Laois/Offaly
L17	Wexford
L18	Waterford
L19	South Tipperary
L20	Carlow/Kilkenny
L21	Cork South
L22	Cork North
L23	Cork West
L24	Kerry
L25	Limerick
L26	Clare
L27	Tipperary North/East Limerick
L28	Galway
L29	Roscommon
L30	Mayo
L31	Sligo/Leitrim
L32	Donegal

The remaining digits of the identifier will then commence with the year (i.e. 2011 followed by a unique 4 digit number given by the LHO (e.g. 0001 for the first non-Irish hospital birth notified each year, 0002 for the second).

These will be allocated by the DPHN (or her/his nominee) and recorded in a file held locally (either electronically or in a manual file).

- Therefore the third baby recorded in such a manner in Dublin Area 5 in 2012 would have the following UPI **L05 20120003**
- The third baby recorded in such a manner in Longford/Westmeath in 2012 would have the following UPI **L15 20120003**

Appendix X: DPHN/CHO/LHO Birth and Newborn Bloodspot Screening Register

Record Keeping Instruction for Birth Register and NBS Register

- The Director of Public Health Nursing is responsible for monitoring Newborn Bloodspot Screening carried out in the Community and for providing a fail-safe procedure to check that all babies have been screened (Hospital or Community) and have received results.

- The Immunisation/PHR/Child Health ICT System is the electronic birth register for Community Healthcare Organisation (CHO) / Local Health Office (LHO).
  - ✓ Some CHOs have an Immunisation/PHR System as Birth Register for all babies who reside in the CHO/designated area.
  - ✓ Some CHOs have a customised in-house Information Technology System that is used as above.
  - ✓ Some CHOs have a manual system which is used as a birth register. For those who have a manual system they should progress to linking to the Immunisation/PHR system as a birth register.

- A NBS Register logs all babies who reside in the CHO/designated (LHO) who have a NBS Sample completed in the Community and the Hospital.

- The NBS Register is located in a designated office (Child Health Office/DPHN/ADPHN Office). In some CHOs there is a Shared Folder ICT arrangement which allows the ADPHN and Designated Officer in Administration to access data for birth notification.

- The NBS Register is usually in the form of an Excel spreadsheet. Data can be extracted from the Immunisation/PHR system where available to create the basis for a NBS Register.

- An excel spreadsheet facilitates counts and has the following datasets:

  - Demographic details: These details are exported from the Immunisation/PHR/Child Health ICT system:
    - ✓ Baby and mother identifying demographics (name/s, address, gender, UPI and/ or Immunisation/PHR/Child Health ICT system number etc)
    - ✓ <u>Baby DOB</u>

  - Details of Date of Collection opt, out and ineligibility: These details are entered by the local office:
    - ✓ <u>Ineligible for NBS on Day 4 after DOB</u> (*Dropdown with choices – see ineligibility below*).
    - ✓ <u>Opt-out</u> (*Dropdown with choices Yes/No).*
    - ✓ Initial sample collection date (dd/mm/yy).
    - ✓ Sample taker and location of sample taken e.g. hospital or community.
    - ✓ Date and detail of repeat tests required (*Dropdown with choices Yes/No, date of repeat request* (dd/mm/yy)*, sample taker detail)*

  - Details of Results:
    - ✓ <u>Date first result received (dd/mm/yy)</u> – this is the date of first result on any of the 6

conditions tested (N.B. It excludes the Beutler test and report that sample is unsuitable).

- ✓ Result of first sample for the 6 conditions (N=Normal; R=Repeat requested RTF =Result to Follow and then enter N or R).
- ✓ Notes: reason for repeat results required.
- ✓ Result by Day 18 (this is populated by a formula which subtracts DOB from Date first result received)
- ✓ Result by Day 18: number of babies with Complete and Incomplete results by day 18.
- ✓ Result by Day 28: number of babies where *Incomplete* first results at Day 18 are completed by day 28. Babies with incomplete results will be tracked until screening is closed and either conditions are not suspected or the baby is referred for treatment.
- ✓ Date of Repeat Results received
- ✓ Result of Repeat Results (e.g. CHT=N; GAL= N)
- ✓ Overall result (*Dropdown with choices All Not suspected/Refer)*- if any further detail / explanation required re follow up of delayed NBS screening, etc)

- Details of Ineligibility: Enter Yes where applicable.
  - ✓ Babies moved into CHO/LHO within 3 days.
  - ✓ Babies moved out of CHO/LHO after 5 days.
  - ✓ Babies who have died.
  - ✓ Babies born in another jurisdiction.
- Details for single record sent to PHN for CHR: Enter Yes.
  (The Core fields required for KPI calculation are **underlined**).

- This data needs to be compiled on a regular basis (enter data daily and do checks weekly).

- Sample Results are accessed via eReports by the Authorised Users allocated by the National Newborn Bloodspot Screening Laboratory. Sample Results are retained for 60 days and are archived thereafter by the NNBSL.

- Sample Results are checked against the Birth Register to ensure all babies residing in the designated LHO area have been screened and relevant parties have received test results.

- If no results are received for the baby who is entered in the NBS Register, the National Newborn Bloodspot Screening Laboratory is contacted by the ADPHN/Designated Officer. The ADPHN ascertains the location of the baby:
  - ✓ Where a baby's details have changed, the designated officer amends the NBS Register and informs the Immunisation/PHR/Child Health ICT system, to ensure both databases are aligned.
  - ✓ Where the baby has moved into the area, the designated officer notes the eligibility of the baby for NBSP and amends the baby's details on the NBS Register.
  - ✓ Where the baby has moved out of the area, the designated officer amends the NBS Register and informs the Immunisation/PHR/Child Health ICT system and relevant CHO, DPHN/LHO.
  - ✓ Where the baby has had the NBS screening in another jurisdiction, the baby's details remain on the NBS Register and the baby is entered as ineligible.
  - ✓ Where the baby has died or miscarried, the baby's details remain on the NBS

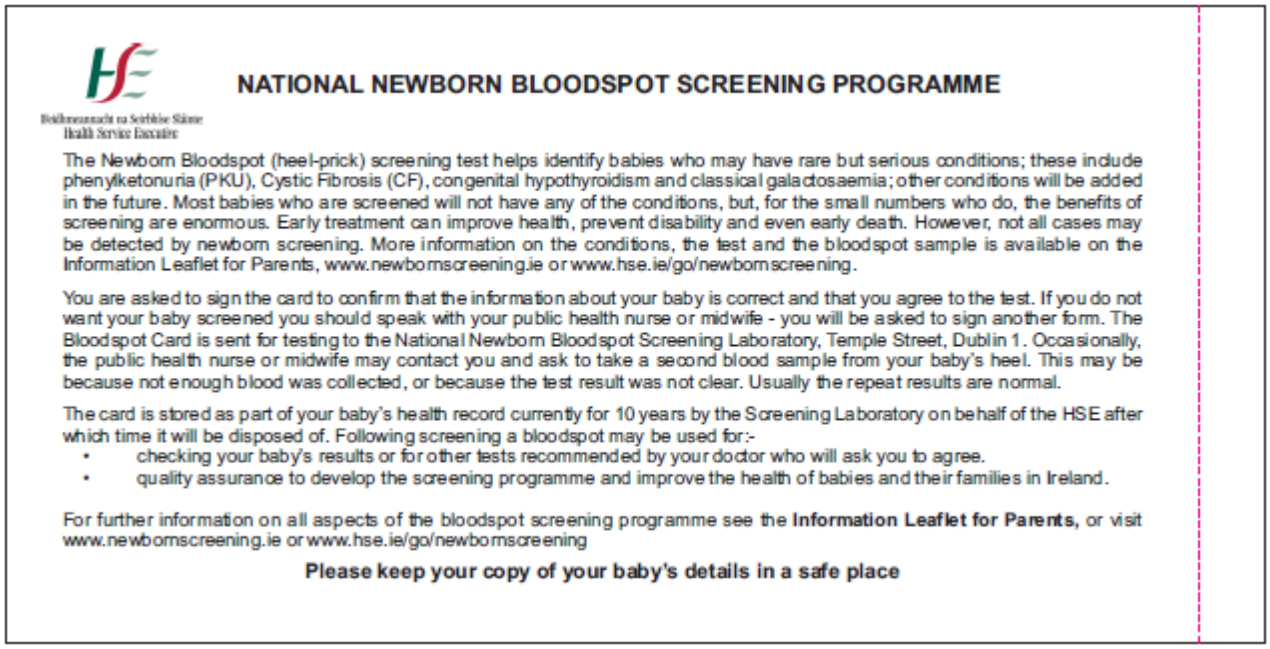
Register and the baby is entered as ineligible.

- The register is checked weekly to ensure all babies have received a result by day 18.

- If the RPHN /RGN receives a request directly (i.e. other than through Liaison/ADPHN) for the Newborn Screening Sample to be taken on a child in her PCT area she will notify the DPHN/ADPHN so that the details of the child can be entered in the NBS Register.

It is noted that there is ongoing work to develop a National Immunisation and Child Health Information System which will stream line much of this work.

Appendix XI: Newborn Bloodspot Screening Card Parent Information Copy



Healthcare Service Executive

NATIONAL NEWBORN BLOODSPOT SCREENING PROGRAMME

The Newborn Bloodspot (heel-prick) screening test helps identify babies who may have rare but serious conditions; these include phenylketonuria (PKU), Cystic Fibrosis (CF), congenital hypothyroidism and classical galactosaemia; other conditions will be added in the future. Most babies who are screened will not have any of the conditions, but, for the small numbers who do, the benefits of screening are enormous. Early treatment can improve health, prevent disability and even early death. However, not all cases may be detected by newborn screening. More information on the conditions, the test and the bloodspot sample is available on the Information Leaflet for Parents, www.newbornscreening.ie or www.hse.ie/go/newbornscreening.

You are asked to sign the card to confirm that the information about your baby is correct and that you agree to the test. If you do not want your baby screened you should speak with your public health nurse or midwife - you will be asked to sign another form. The Bloodspot Card is sent for testing to the National Newborn Bloodspot Screening Laboratory, Temple Street, Dublin 1. Occasionally, the public health nurse or midwife may contact you and ask to take a second blood sample from your baby's heel. This may be because not enough blood was collected, or because the test result was not clear. Usually the repeat results are normal.

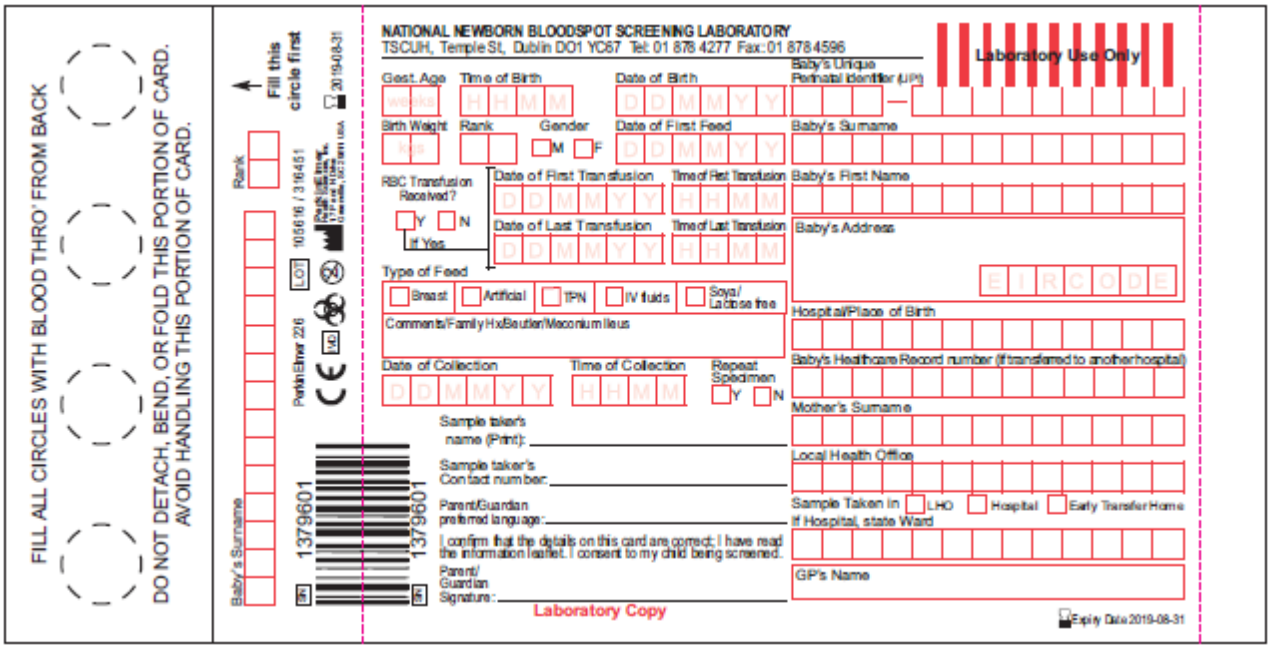
The card is stored as part of your baby's health record currently for 10 years by the Screening Laboratory on behalf of the HSE after which time it will be disposed of. Following screening a bloodspot may be used for:-

- checking your baby's results or for other tests recommended by your doctor who will ask you to agree.
- quality assurance to develop the screening programme and improve the health of babies and their families in Ireland.

For further information on all aspects of the bloodspot screening programme see the **Information Leaflet for Parents,** or visit www.newbornscreening.ie or www.hse.ie/go/newbornscreening

Please keep your copy of your baby's details in a safe place

Appendix XII: Newborn Bloodspot Screening Card



Appendix XIII: National Newborn Bloodspot Screening Programme Opt-Out Form

Seirbhís Sláinte Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

NATIONAL NEWBORN BLOODSPOT SCREENING PROGRAMME

OPT-OUT FORM

Baby's Unique Perinatal Identifier (UPI): ___ - _____ Gender: *(Please circle)* M F

Baby's Surname: _____ Gest Age: *(weeks)* ____

Baby's First Name: _____ Rank: ____

Baby's Address: _____ Hospital/Birth Place _____

Mother's Surname: _____

Time of Birth: H H M M Date of Birth: D D M M Y Y Birth Weight *(kgs)*: ___ - ____

RBC Transfusion Received? Y / N *If Yes* Date of First Transfusion: D D M M Y Y

Time of First Transfusion: H H M M

Date of First Feed: D D M M Y Y Local Health Office: _____

- I _____ being the parent/guardian of Baby _____ do not consent to allow the Newborn Bloodspot Screening Test (Heel-Prick) to be carried out on my baby.
- I have read the information leaflet on Newborn Bloodspot Screening and the test has also been explained to me.
- I fully understand the importance of the decision that I am taking by not allowing my baby to be tested.
- I understand that not detecting or treating one of the conditions, should baby have one, may result in severe intellectual or physical disability which could require long term care or result in premature death.

Signed (Parent/Guardian): _____

Full Name (PRINT): _____ Date: _____

Signed by potential sample taker: _____ Date: _____

Position/Job Title: _____ Health Area Office: _____

OFFICIAL USE ONLY

6 copies of the completed form, signed by parent/guardian and sample taker should be made. A copy to be given to the parent/guardian and a copy kept by the sample taker.

Copies are to be posted to each of the following:- • The Director of Nursing/Midwifery • Director of Public Health Nursing • National Newborn Bloodspot Screening Laboratory • The baby's General Practitioner

	Director of Nursing/Midwifery	Director of Public Health Nursing	National Newborn Bloodspot Screening Laboratory (NNBSL)	General Practitioner
Name				
Address			NNBSL, Children's University Hospital, Temple Street, Dublin 1	
Date Sent				

Signed *(Sample taker)*: _____ Date: _____

Name *(Block Capitals)*: _____

NOTE: *Parents/Guardians may change their mind but it is their responsibility to inform their GP or PHN*

The Cystic fibrosis screen is not suitable if baby is over six weeks of age

LF-NNS-0116 ***www.newbornscreening.ie***

Appendix XIV: High Risk Screening

***HSE A Practical Guide to Newborn Bloodspot Screening in Ireland 7th Edition* (2018) Page 32-37**

High Risk Screening

N.B. Note relevant family history on screening card and inform metabolic laboratory Temple St 01 8784670/4727

Siblings of a known confirmed positive case

Phenylketonuria (PKU)
A newborn screening sample (NBS) and a lithium heparin blood sample should be taken between 72-120 hrs. following birth AND another NBS sample on day 10.
If the 72 hrs. following birth is due to fall on a Saturday, then we recommend that the liquid sample is taken and sent to the Metabolic laboratory in Temple St on the Friday morning and laboratory phoned in advance, in order to avoid parental anxiety over the weekend.

Homocystinuria (HCU)
A NBS should be taken between 72 -120 hrs following birth AND a lithium heparin blood sample taken at the same time for plasma methionine, total homocysteine and free homocysteine and be immediately deproteinised. A further lithium heparin blood sample should be taken on day 10 of life and again deproteinised immediately.
N.B. *For advice on sample deproteinisation, contact the Metabolic Laboratory in Temple St.*

Maple Syrup Urine Disease (MSUD)
A lithium heparin blood sample should be taken on **Day 1** after the second feed and then **DAILY** until established on full feeds. Urine should be tested daily for ketones. A routine NBS sample should be taken between 72-120 hrs. to test for the other conditions.
Then a further NBS card or lithium heparin sample at day 10. Staff at the NNBSL and the National Centre for Inherited Metabolic Disorders, Temple Street must be informed, prior to the delivery of the baby.

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) It is very important that any baby at risk of having MCADD due to a relevant family history is tested at the earliest opportunity. The pregnancy should be discussed with the metabolic team in the National Centre for Inherited Metabolic Disorders, Temple St., early in the pregnancy for careful management of birth to minimise the risk of decompensation. Management at birth may depend on the presentation of the previous sibling.

A dried blood spot sample should be collected on a metabolic card at 24–48 hours for dried blood spot Acylcarnitine profile, indicate if baby is on IV fluids, glucose or dextrose and also collect a sample for urinary organic acids (5mls fresh urine, frozen, with no preservative added). Then take a routine sample, between 72-120 hours to test for all the other conditions and a repeat screening card at day 10.

It is extremely important that any baby at risk of having MCADD due to a relevant family history is tested at the earliest opportunity. The pregnancy should be discussed with the Metabolic Clinical Team for careful management of birth to minimise decompensating risk. Management at birth may depend on the presentation of previous sibling.

N.B.: Feeds protocol for high risk (sibling) MCADD screens
It is essential to ensure that the baby maintains a good milk intake until results are available. If baby is well it

should be bottle fed every 3 hours or if breast fed every 2-3 hours during day time and every 3 hours at night (at least 10 minutes on the breast).
Exclusively breast fed babies are particularly at risk in the first few days when the supply of breast milk is poor; top up feeds of expressed breast or formula milk may be necessary until a good milk supply is established. Seek advice from the Metabolic clinical team in Temple St.

If oral feeds are not tolerated, or if the baby is unwell in any way, urgent referral should be made to a Metabolic Paediatrician for review and consideration of IV glucose.

Glutaric aciduria type 1 (GA1)
A dried blood spot sample, taken at 24–48 hours on a metabolic screening card for dried blood spot Acylcarnitine profile and urinary organic acids (5mls fresh urine, frozen with no preservative added). A routine NBS sample should be taken between 72 -120 hours to test for all the other conditions and a repeat screening card at day 10. Feeding routine needs to be established and the baby should continue with oral feeds.

Cystic Fibrosis (CF)
Soon after birth, an EDTA blood sample should be sent directly by the maternity unit/hospital (if parent(s)/legal guardian(s) request/consent), to the Department of Medical Genetics in OLCH, Crumlin, clearly stating the name and DOB of the affected sibling. Take a routine NBS screening sample at 72-120 hrs.
N.B. Blood IRT measurement is unsuitable for CF screen in infants older than six weeks of age.

Congenital Hypothyroidism (CHT)
Thyroid function tests should be performed on day three of life on a lithium heparin/serum blood sample (check local guidelines). The risks are dependent on the type of CHT. For those with dyshormonogenesis the risk is one in four, while those with a family history of thyroid agenesis or dysgenesis the risk is about one in 2,000, depending on the gender of the baby; thyroid agenesis/dysgenesis CHT is more common in girls than in boys.

Classical Galactosaemia (CGAL)
Sibling testing, known family history and Irish Traveller babies including settled travellers

The incidence of Classical Galactosaemia among members of the Irish Travelling community is high at one in 450 births.
To screen for Classical Galactosaemia, a NBS sample should be taken immediately after birth and before any blood transfusion has been given. A cord blood sample is not suitable.

The sample should be sent to the NNBSL for a 'Beutler Test'. This test measures the enzyme activity in red blood cells (this is why a RBC transfusion invalidates the test) and is NOT dependent of feeds. The sample should be clearly marked "FOR BEUTLER TEST". The Beutler assay is not performed as an emergency on-call investigation, unless clinically indicated. However, they are performed on Saturday mornings providing that the sample is received in the NNBSL by 10.00am. Special provision is made for Bank Holidays and long weekends. Two fully saturated circles are required.

N.B.: All *at-risk* babies should be fed with lactose/galactose free feeds (e.g. SOYA feeds) until the result of the Beutler test are known. The routine NBS sample should be taken between 72-120 hours following birth to test for the other conditions.

Baby born to a parent diagnosed with a disorder on newborn bloodspot screening panel
Babies born to a parent with a condition included on the newborn screening panel are at high risk of having the condition, the risk being approximately twice the carrier incidence for the disorder within the Irish population. Irrespective of the condition, these babies should be screened as outlined above as if they were a sibling of a confirmed case for the specific condition.

Family history of a metabolic disorder in extended family
If there is a history of a metabolic disorder within the extended family other than a sibling, this should be clearly stated on the screening card to include the name of the disorder, in such circumstances please contact the NNBSL or NCIMD Temple Street for advice.

Cousin marriages within the Irish Traveller community

Some members of the Irish Traveller community might wish to seek genetic counselling advice; this can be arranged through the Department of Medical Genetics, OLCH, Crumlin.

Maternal phenylketonuria

Phenylalanine is actively transported across the placenta; the blood level in the foetus is about twice that of the mother. Therefore, women who themselves have PKU, should plan conception so that their condition is under optimal control at the time of conception. Regular and frequent monitoring of blood levels of phenylalanine and tyrosine are required throughout pregnancy in order to safe-guard the well-being of their foetus. Following the birth of the baby a liquid sample should be taken from the baby at the same time as the newborn screening sample (72-120 hrs.) and a repeat screening sample on day 10 of life.

Babies born to immigrant parents or refugees

Arriving before NBS has been performed in country of birth
All babies and infants of immigrant parent(s)/legal guardians, up to one year of age, who arrive in Ireland before any newborn screening test has been performed, should be screened for all the conditions on the Irish panel. Screening for CF by measuring blood IRT is not reliable for any infants over six weeks of age.

Arriving after NBS screen has been performed in country of birth
The sample taker should be aware that many countries including Northern Irelands newborn screening programme does not include screening for Classical Galactosaemia. If there is clinical concern of Classical Galactosaemia, then the Beutler test should be requested on a dried bloodspot. The full screening panel (excluding CF if baby over six weeks of age) can be performed on babies under one year of age.

Decision to screen is a local clinical decision, dependent on family history and country of origin of parents. Often a venous sample is taken and spotted onto the card as it may not be possible to take a heel prick sample, it is imperative that this sample is whole blood and has not been collected in a tube with any preservative (such as EDTA, lithium heparin etc.). For children greater than one year, screening can be performed (excluding CF screen) if there is concern raised by the Paediatrician.

If there is no documented evidence in the child's medical records that screening was performed please contact the NNBSL who may have a record and can advise.

The NNBSL on occasion performs TSH analysis on dried blood spot samples from
Down Syndrome patients, as it is often less traumatic than a venous sample.

Babies presenting with meconium ileus at birth
Meconium ileus is a common complication in babies with CF, occurring in about 18% of all CF babies born in Ireland. Not all these infants present with a raised blood IRT (i.e. a positive CF screen) from the NBS screen. Therefore, CF should be considered in all babies who present with meconium ileus within the first days of life. An ETDA sample should be collected and sent directly by the maternity unit/hospital to the Department of Medical Genetics, OLCH, Crumlin for CF mutation analysis, giving full clinical information.
The routine NBS sample should be taken at 72- 120 hours to screen for the other conditions

Procedure for infants over six weeks who have missed newborn screening for Cystic Fibrosis
Children who have missed the 72-120 hours window for newborn bloodspot screening (NBS) for logistical reasons, (clinical condition, lost screening card, delay in receipt in laboratory, etc.) can have their screen completed without delay when issues identified.

Immunoreactive trypsinogen (IRT) the screen for cystic fibrosis (CF) is not suitable if the infant is over six weeks of age. This applies to premature infants as well as term infants. Beyond six weeks of age the IRT value is not interpretable and a bloodspot sample is invalid for the purpose of CF screening. Therefore, for children greater than six weeks of age who were missed or there was a problem with the sample, or the testing, such that no result for CF NBS is available, a sweat test should be performed to out rule CF.

The sweat test should be performed in one of the six specialist CF centres involved in accepting children for assessment as per the CF NBS programme. The following steps should be undertaken;

1. The individual aware of, or concerned that, a child has not been screened for CF as part of the CF NBS programme should inform the National Newborn Bloodspot Screening laboratory (NNBSL) in the Children's University Hospital, Temple St, Dublin.
2. The NNBSL should collect the relevant details of the infant involved, and the details of why screening was missed, and then contact the specialist CF centre nearest the baby's geographic catchment area to request a sweat test.
3. Contact with the specialist CF Centre should use the existing lines of communication for the CF NBS programme; i.e. a telephone call to the CF clinical nurse specialist (CNS) followed by faxed written details. This will be carried out by the NNBS laboratory.
4. The CF CNS will organise a sweat test and review by a CF Consultant as per the usual procedure for the CF NBS programme.
5. The specialist CF centre will provide the results to the parents/legal guardians and NNBSL as soon as they are available.
6. The NNBSL will inform the individual who highlighted the case that the infant has been assessed and will inform the maternity unit and the Director of Public Health Nursing in that area.

The procedure as described above does not apply to infants who had newborn bloodspot screening performed in another country which did not include CF NBS, and then moved to Ireland at a later date. These children should not be screened for CF, but should have a sweat test performed only if clinically indicated, such as if they develop clinical features, or have a strong family history of CF.
If unable to perform a sweat test, for example if skin condition deems it unsuitable, genetic analysis to be offered following parental consent.

Appendix XV: National Newborn Bloodspot Screening Laboratory Contact Details

Contact Details	
Children's University Hospital **Temple Street, Dublin 1, D01 YC67**	(01) 878 4200
Newborn Screening Laboratories	
Enquiries	(01) 878 4577
Fax	(01) 878 4596
Email Address	info.newbornscreening@cuh.ie
Director Dr. Ingrid Borovickova	(01) 878 4266
Chief Medical Scientist (Ms Loretta O' Grady)	(01) 878 4277
Clinical Liaison Officer (Ms Olivia Walsh)	(01) 892 1804
National Centre for Inherited Metabolic Disorders	
Enquiries	(01) 878 4317
Laboratory Opening Hours	
Monday to Friday Analysis including Beutler samples and reporting of results	**09:00-17:00**
Saturday Morning Analysis including Beutler samples and reporting of results. Beutler samples must be in the NNBSL before 10:00	**09:00-12:00**
Christmas and Easter Holidays:	Opening hours will be circulated well in advance and/or posted on the website

Appendix XVI: Newborn Bloodspot Screening Card Checklist for Sending Samples Together

Newborn Bloodspot Screening Card Checklist

IMPORTANT

Ensure samples are dry, envelopes are sealed and no lancets enclosed

Please complete this form and list each baby's name and UPI as written on the screening cards.

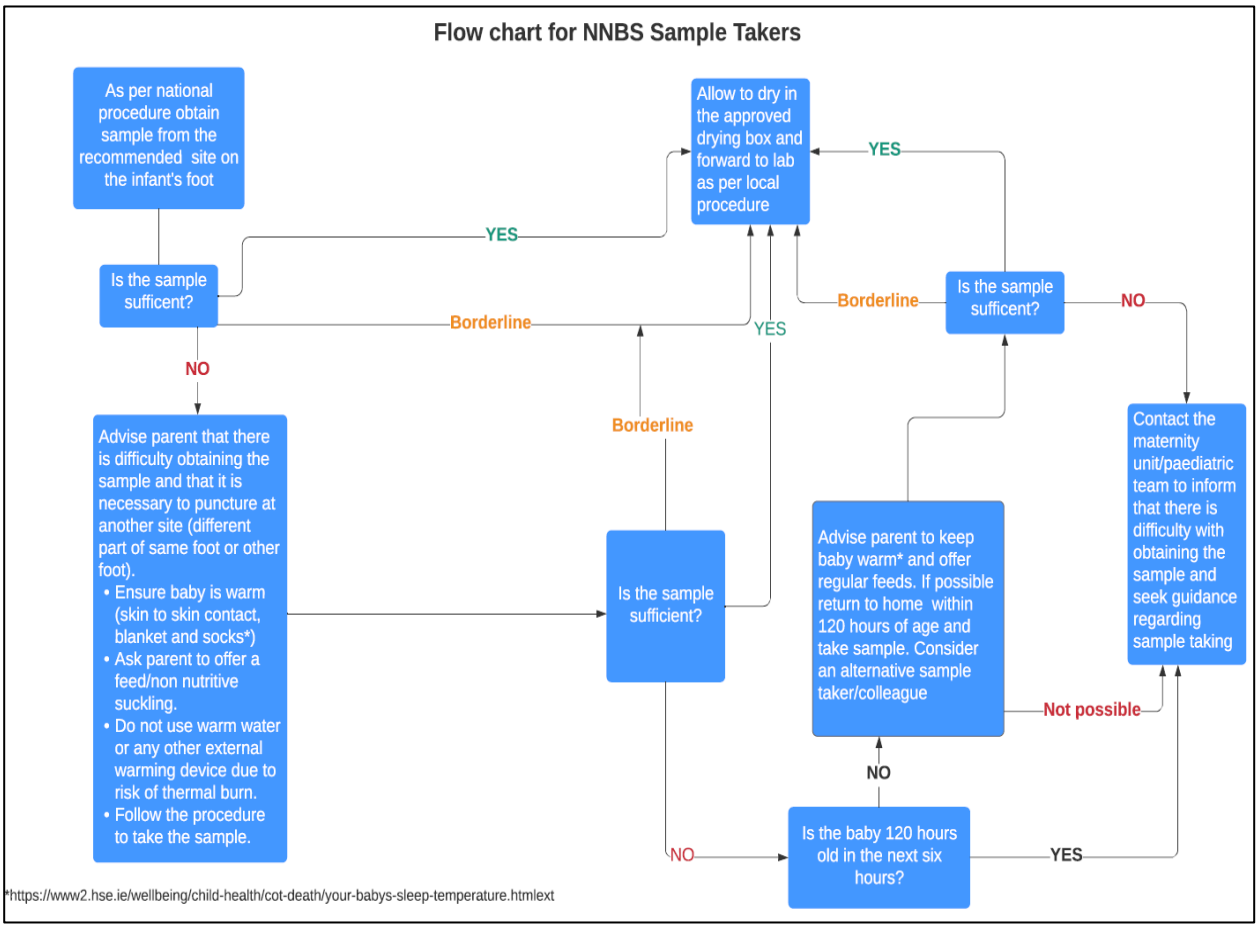
This form should be included in all envelopes containing more than one screening card (maximum of 3 Samples)

The National Newborn Bloodspot Screening Laboratory
Children's University Hospital, Temple Street, Dublin 1, D01 YC67
Ph: 01 8784610/4277

Location:		**Contact No.:**
Total number of cards:	**Date Posted:**	**Checked by:**

Baby's Surname (as on card)	**Baby's UPI (as on card)**	**Additional Comment** *if relevant*

Appendix XVII Flow chart for Newborn Bloodspot Screening Sample Takers



Appendix XVIII: Transport/Drying Box – Procedure for Transporting Samples

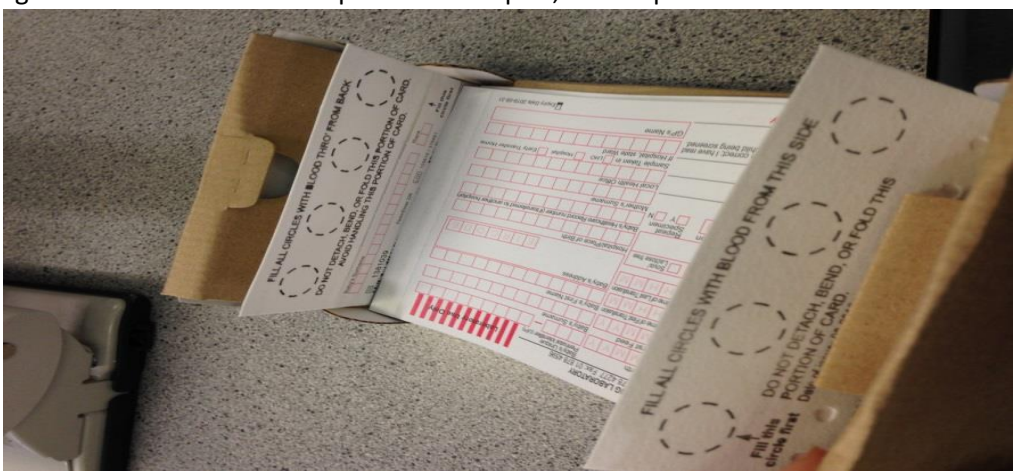
Transport/Drying Boxes

- The National Newborn Bloodspot Screening Laboratory has designed drying/transport boxes in conjunction with Mega Pak to facilitate the transport of NBSCs from the baby's home to the PHNs car in a safe manner. Once the bloodspot has dried, the NBSC should be removed from the box and packaged according to the regulations. These drying boxes can contain two NBSCs.

- The boxes are reusable. However, if they become contaminated with blood they should be disposed of either by incineration or through the accepted procedure for disposable of hazardous waste.

- These boxes can be ordered directly from Mega Pak Ltd by the Public Health Nursing Department. The minimum order is 150 boxes, flat packed in batches of 50 from Mega Pak Ltd (Irish Office), 16 Highfield Green, Swords, Co Dublin. Tel: 01 8402063, email megapakireland@eircom.net, website www.mega-pak.com

Drying box for two NBS cards: top and tail samples, blood spots should not come in contact



Procedure for transporting samples

The sender of NBSCs by registered post or by courier is responsible for ensuring that the packaging and transportation of the sample complies with current transport regulations regarding Health and Safety as laid down in the European Directive (ADR 2015) Packaging Regulations P650.

Dried bloodspots must be packaged appropriately. NNBSL recommends that once the blood has dried, the NBSC should be inserted into a water-resistant, tear-proof Tyvek® envelope or equivalent. The yellow fluorescent address label should be fixed to the outer envelope.

Pre-printed registered envelopes

Pre-printed plastic envelopes may be purchased directly from An Post by e-mailing both Brian Beehan (brian.beehan@anpost.ie) and Noreen Hudson (noreen.hudson@anpost.ie)

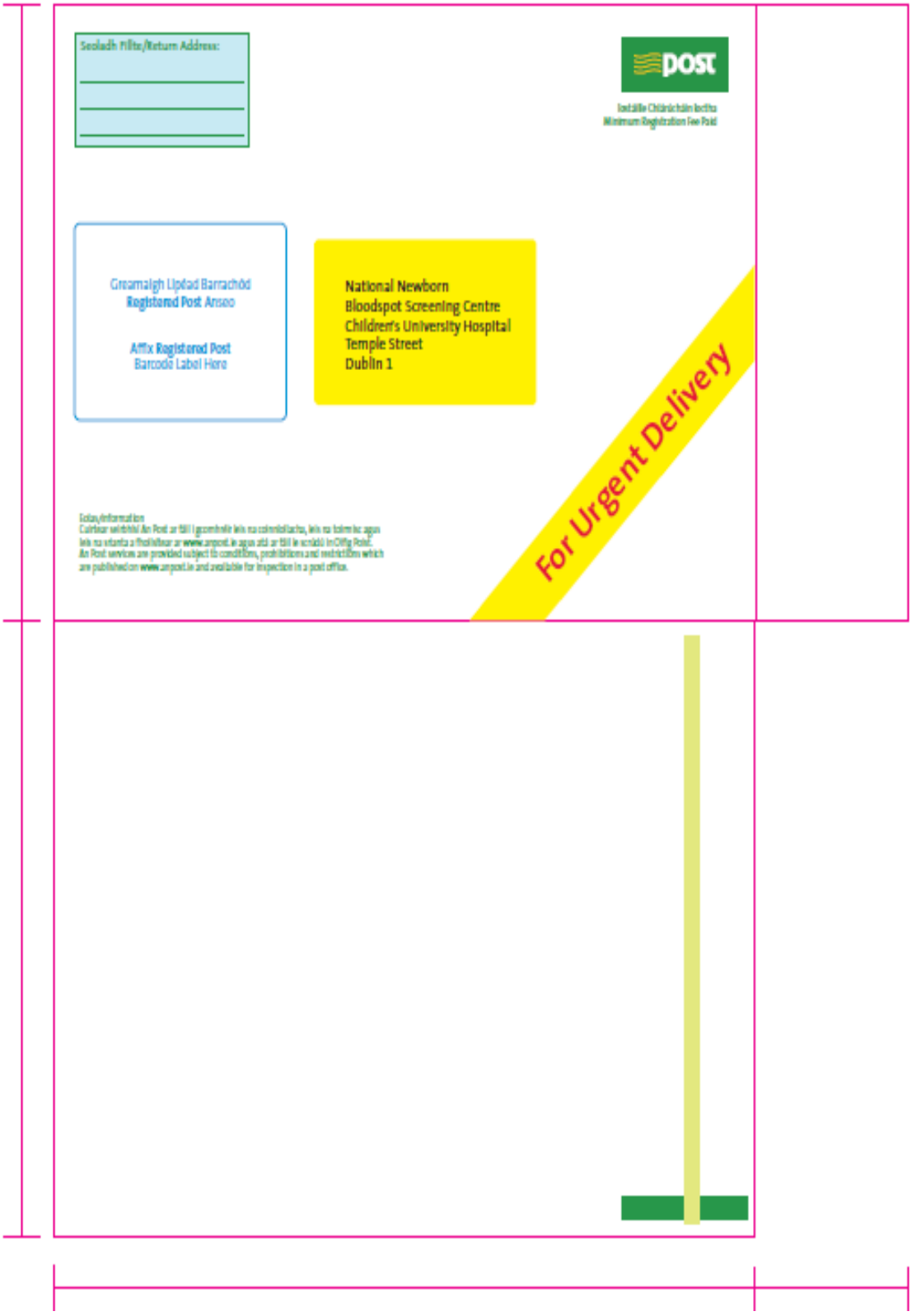
Please send an email for the envelopes to both persons named above (in case one is on leave), stating the amount of envelopes required and quoting an HSE purchase order number. Envelopes retail @ €6.85 each and come in packs of 10 @ €68.50 per pack. A Pro Forma invoice, with the HSE purchase order number as reference, will then be returned to enable payment by EFT. Once the EFT payment has reached An Post's bank account the Philatelic Section will dispatch the order.

Responsibility of sender

If more than one NBSC is put in an envelope, they should be placed at 180° to each other (i.e. the bloodspots should not overlap and therefore not touch). The sender should state in writing how many NBS cards are in each envelope and include a list of the names of the babies on a separate page, a sample checklist is available to download from www.newbornscreening.ie

NBSCs, appropriately packaged, should be sent daily and not batched. It is the responsibility of the sender to dispatch the sample as soon as possible after collection, either by registered post or by courier. It is not appropriate to put the package into the post knowing that there may be a delay in it arriving at the NNBSL due to either a postal dispute (local or national) or over the Christmas period when the post is delayed. Alternative arrangements should be made by maternity hospitals/units and LHOs to ensure NBSCs are dispatched to NNBSL without delay. Parents should never be asked to post or deliver NBSCs to the NNBSL. This is the responsibility of the sample taker.

Appendix XIX: Waterproof and Tear Proof Envelope for Registered Post



Appendix XX: Primary Childhood Immunisation/GP Forename Card

Confidential

Notification to Primary Care Services from the Public Health Nurse

Primary Childhood Immunisation Scheme

Child's Register Name: …… Mother' Name:……..
(NB as per Birth certificate)

Child's Date of Birth: …….. Mother' Forename:……

Male/Female Mother' Maiden Name:……..

Child's PPS No.: ……..…… Mother' PPS No.:……

Mother' Marital Status:
Married ☐ Single ☐
Widowed ☐ Separated ☐ Divorced ☐

Discharge Address (If different from permanent address): ……..

……..

Parents Daytime Contact Number:……..

DED (For Official Use) …… Local Health Office (LHO)

PHN Area:…….. PHN Name:…….

Detail of Birth:
Time of Birth…….. Place where NBS Sample completed
Hospital ☐ Primary Care Services ☐

Sampler Taker's Name:……. Date Sample Taken……

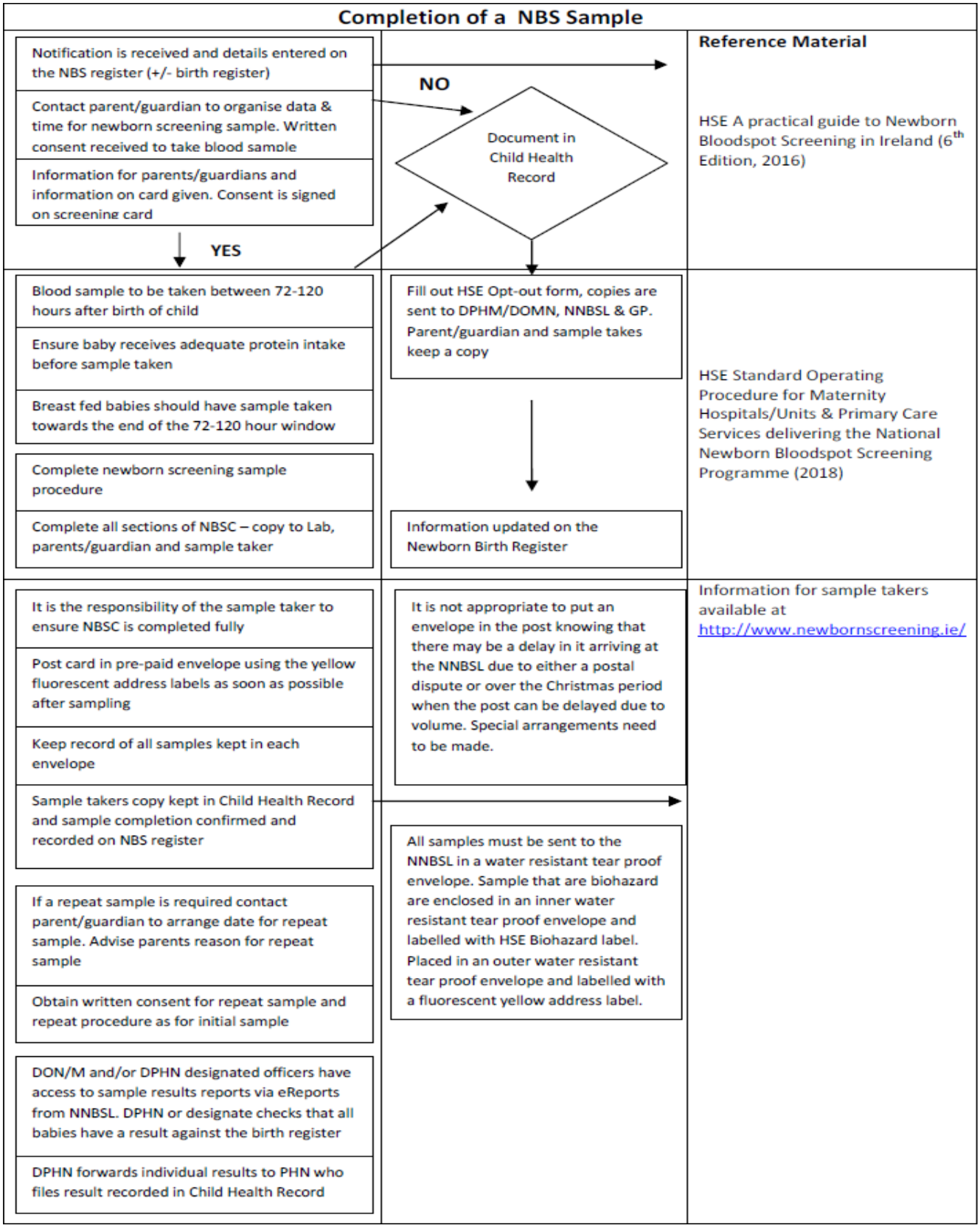
Date sample Posted:……

GP Nominated for vaccination

GP Full Name:……..GP Contact Number: ……..

GP Address:……

Appendix XXI: Flow Chart Checklist for Newborn Bloodspot Screening Sample Collection



Appendix XXII: Temple Street Children's University Hospital – Copy Result Request Form

No.	Baby Surname	Mother Surname	DOB	Baby Forename	Mother Forename	Address	Hospital of Birth	Unique Identifier (UPI)	NNBSL Office Use/ Comments
1									
2									
3									
4									
5									
6									

N.B.: Please ensure e-reports have been checked prior to sending this form requesting copy reports.

Requested by: _____ **Address:** _____

_____ **Tel No.:** _____

Issued by (NNBSL use only): _____ **Date** (NNBSL use only):_____

Appendix XXIII: Informing NNBSL of reports received that do not belong to CHO/LHO Area

Please complete details of this form:
For babies on whom you have received reports who do not belong to your Area
Return the Form to NNBSL. Please indicate correct LHO (if known).
Please email or FAX (on headed paper) to NNBSL as soon as possible

Location (LHO):	**Date:**
Reported By: (Print name and provide signature)	**Position:**

UPI	Lab Number	Baby's Surname	Baby's First Name	Date of Birth	Mothers Surname	Old Address	New Address	Correct LHO (if known)

Comment:

Appendix XXIV: Individual Sample Results – Print Out

National Newborn Bloodspot Screening Laboratory

An INAB accredited testing Laboratory Reg. No.224MT
Temple Street Children's University Hospital, Dublin 1
Director: [redacted]
Chief Medical Scientist:
Tel: 01 8784 277 Fax: 01 8784 596 Email: info.newbornscreening@cuh.ie: www.newbornscreening.ie

Feidhmeannacht na Seirbhíse Sláinte
Health Service Executive

Newborn Bloodspot Screening Report **Report date:** 01/08/2017

Location: **Paediatric OPD Rotunda Hospital (RH)**

Baby Details

Name:	**TEST REPORT, TEST**	**UPI:**	**932-123456**	**Sex:**	
Mother's Surname:	TEST				
Address:	123 NEW STREET, TEMPLE ST, D01 YC67				
DOB:	**27/07/2017**	**Lab number:**	17-217001		
Date of collection:	01/08/2017	**Time of collection:**	09:00		
Date received:	01/08/2017	**Sample type:**	Initial		

Sample taker name: JOE BLOGGS **Contact number:** 01 8784612
Location sample taken:

Screening results

Condition	Result	Result Codes NNBSL	Result Codes HSE
PKU (Phenylketonuria)	Not Suspected	N	2
HCU (Homocystinuria)	Not Suspected	N	2
MSUD (Maple Syrup Urine Disease)	Not Suspected	N	2
GAL (Classical Galactosaemia)	Not Suspected	N	2
CHT (Congenital Hypothyroidism)	Not Suspected	N	2
CF (Cystic Fibrosis)	Not Suspected	N	2

Report authorized by Loretta O'Grady 01/08/2017 15:46

Page 1 of 1 Report version 1.3

[redacted]

Appendix XXV: Query Positive Results

HSE A Practical Guide to Newborn Bloodspot Screening in Ireland 7th Edition (2018, Page 52-56)

Referral procedures for specific conditions are set out below:

Query Positive Cases for PKU

The Clinical Liaison Officer in the NNBSL will book a bed for admission to Temple Street Children's University Hospital and liaise with the on-call metabolic team. The designated liaison nurse in the maternity unit/hospital will be contacted and asked to:

- Arrange for the baby to attend Temple Street Children's University Hospital under the care of the on-call Metabolic Paediatrician;
- Give the contact number of the Director of the NNBSL (or deputy) to the parent(s)/legal guardian(s), and invite them to make contact for more information, if they wish to do so.

If the initial blood phenylalanine level is high then it is likely that the baby will be kept in hospital for a number of days until the level has fallen. During this time the baby will be started on lifelong dietary treatment and the parents will receive instruction on the monitoring and dietary management of their baby. This should be the only time that the baby will be admitted to hospital for the specific management of PKU.

Babies with a milder variant of the condition may be referred to the outpatient clinic. This information will be clearly given to the designated liaison nurse at the time of the initial contact.

Query Positive Cases for MSUD

The Director of the NNBSL or deputy will discuss the case with the on-call Metabolic Paediatrician. He/She will then either contact the designated liaison nurse or the Paediatric Registrar in the maternity unit/hospital directly, and arrange for the baby to be admitted as a matter of urgency, either to the local Special Care Baby Unit or directly to Temple Street Children's University Hospital.

If the baby is to be admitted to:

- ***the local SCBU***
  - Explain to the parents what disorder the baby is suspected of having
  - On admission, examine the baby in detail and check the urine for the presence of ketones;
  - Arrange for 1.3mL of whole blood collected into a lithium heparin tube for plasma branch chain amino acids to be sent immediately to the Metabolic Laboratory, Temple Street.

  Contact the on-call Metabolic Paediatrician at Temple Street for further advice on management.

- ***Temple Street Children's University Hospital***
  - Parent(s)/legal guardian(s) must be informed that the baby will be admitted to hospital until the results of tests are known. If the test is positive, the baby will remain in hospital until the Metabolic Paediatricians are satisfied that the baby's condition is under control and that the parents will be able to cope at home.

Query Positive Cases for HCU

The designated liaison nurse will be asked to:

- Locate the baby and parent(s)/legal guardian(s) and explain to them why a blood sample is required and what disorder the baby is suspected of having
- Arrange for the baby to have a blood sample taken (1.3 mL of whole blood collected into a lithium heparin tube) for methionine, total and free homocysteine and liver functions tests.

  NB. The sample for free homocysteine must be de-proteinised immediately by the local laboratory staff; instructions will be given over the telephone.

  - If the plasma total and free homocysteine are raised and support the diagnosis, arrangements will be made for the parent(s)/legal guardian(s) to attend the metabolic outpatient at Temple Street Children's University Hospital.
  - If the plasma methionine remains elevated but the total and free homocysteine are not raised and the baby is clinically well, further advice will be given to repeat the test. The baby will be followed up in the metabolic outpatient at Temple Street Children's University Hospital.

Query Positive Cases for Classical Galactosaemia

The Director, Clinical Liaison officer or deputy will contact the Paediatric Registrar in the maternity unit/hospital to arrange immediate admission to the local Paediatric unit. They will be asked to:

- Locate the baby and parent(s)/legal guardian(s) as a matter of urgency;
- Explain to the parents the nature of the condition and to bring their baby directly into the local paediatric unit where the baby will be admitted to hospital for further investigations.

On admission, all lactose and galactose containing feeds including breast milk should be replaced by soya-based feeds (e.g. wysoy). The baby should be examined and the following investigations performed:

- Liver function tests
- Coagulation screen
- Blood cultures (to exclude, for example, *E coli* septicaemia)
- Repeat screening card

As soon as the results of the investigations are available the local clinicians should either contact the Director of the NNBSL or the on-call Metabolic Paediatrician at Temple Street Children's University Hospital to discuss further action.

Query positive cases for MCADD

The Director, Clinical Liaison officer or deputy will contact the Paediatric Registrar in the maternity unit/hospital. They will be asked to:

- Locate the baby and parent(s)/legal guardian(s) as a matter of urgency;
- Explain the nature of the condition suspected and to bring their baby directly into the local paediatric unit for assessment and further investigations.

All babies with an MCADD suspected screening result should be referred to the Metabolic Unit Temple St. on

the same day the screening result is available. The following tests should be performed: urine organic acids and DBS acylcarnitines and the following may be considered: blood glucose, liver function tests, ammonia and CK.

N.B.: Feeds protocol for MCADD screen positives

It is essential to ensure that the baby maintains a good milk intake until results are available. If baby is well it should be bottle fed every 3 hours or if breast fed every

2-3 hours during day time and every 3 hours at night (at least 10 minutes on the breast).

Exclusively breast fed babies are particularly at risk in the first few days when the supply of breast milk is poor; top up feeds of expressed breast or formula milk may be necessary until a good milk supply is established. Seek advice from the Metabolic clinical team in Temple St.

If oral feeds are not tolerated, or if the baby is unwell in any way, urgent referral should be made to a Metabolic Paediatrician for review and consideration of IV glucose.

Query positive cases for GA1

The Director, Clinical Liaison officer or deputy will contact the Paediatric Registrar in the maternity unit/hospital. They will be asked to:

- Locate the baby and parent(s)/legal guardian(s)
- Explain the nature of the condition suspected and to bring their baby directly into the local paediatric unit or Temple St. for assessment.
  If advised by Metabolic Consultant the baby may be admitted to hospital following further investigations.

All babies with a GA1 suspected screening result should be referred to the Metabolic unit Temple St. on the same day the screening result is available. The following tests should be performed: urine organic acids and DBS Acylcarnitine profile. Renal and liver profile may be considered.

N.B.: Feeds protocol for GA1 screen positives

Feeding routine needs to be established and the baby must continue with regular feeding. If oral feeds are not tolerated, or if the baby is unwell in any way, urgent referral should be made to a Metabolic Paediatrician for review and consideration of IV glucose.

Query Positive Cases for Cystic Fibrosis

The results of the screening test will be available by approximately the third week of life:

- The Clinical Liaison Officer will contact the designated liaison nurse in the maternity unit/hospital to obtain the parent(s)/legal Guardian(s) contact details and any relevant clinical information. She/he will then contact the CF Nurse Specialists in the appropriate HSE designated paediatric CF Centres, to give them the full contact details, relevant clinical information and the results of the mutational screen.
- The CF nurse specialist will book a sweat test appointment, and then contact the parent(s)/legal guardian(s) to arrange for the baby to attend the nearest CF centre the following day.

On arrival, the parents will be fully informed as to what will happen; the baby will have a sweat test, the results of which should be available by early afternoon on the same day if sufficient sweat is collected. Depending on the results of the sweat test, the parents will be informed that their baby has CF or is a carrier of the condition. If the baby is considered to be a carrier, and therefore unlikely to have CF, the parents will be referred for

genetic counselling.

HSE Designated Paediatric CF Centres

- Dublin North: Temple Street Children's University Hospital
- Dublin South two locations:
  Our Lady's Children's Hospital, Crumlin
  National Children's Hospital (AMNCH), Tallaght
- Cork: Cork University Hospital
- Limerick: University Hospital Limerick
- Galway: University College Hospital, Galway

Query Positive Cases for Congenital Hypothyroidism

The procedure may vary slightly depending on the age of the baby, the degree of elevation of the blood TSH level and referral hospital. However, clear instructions will be given. The designated liaison nurse will be asked to:

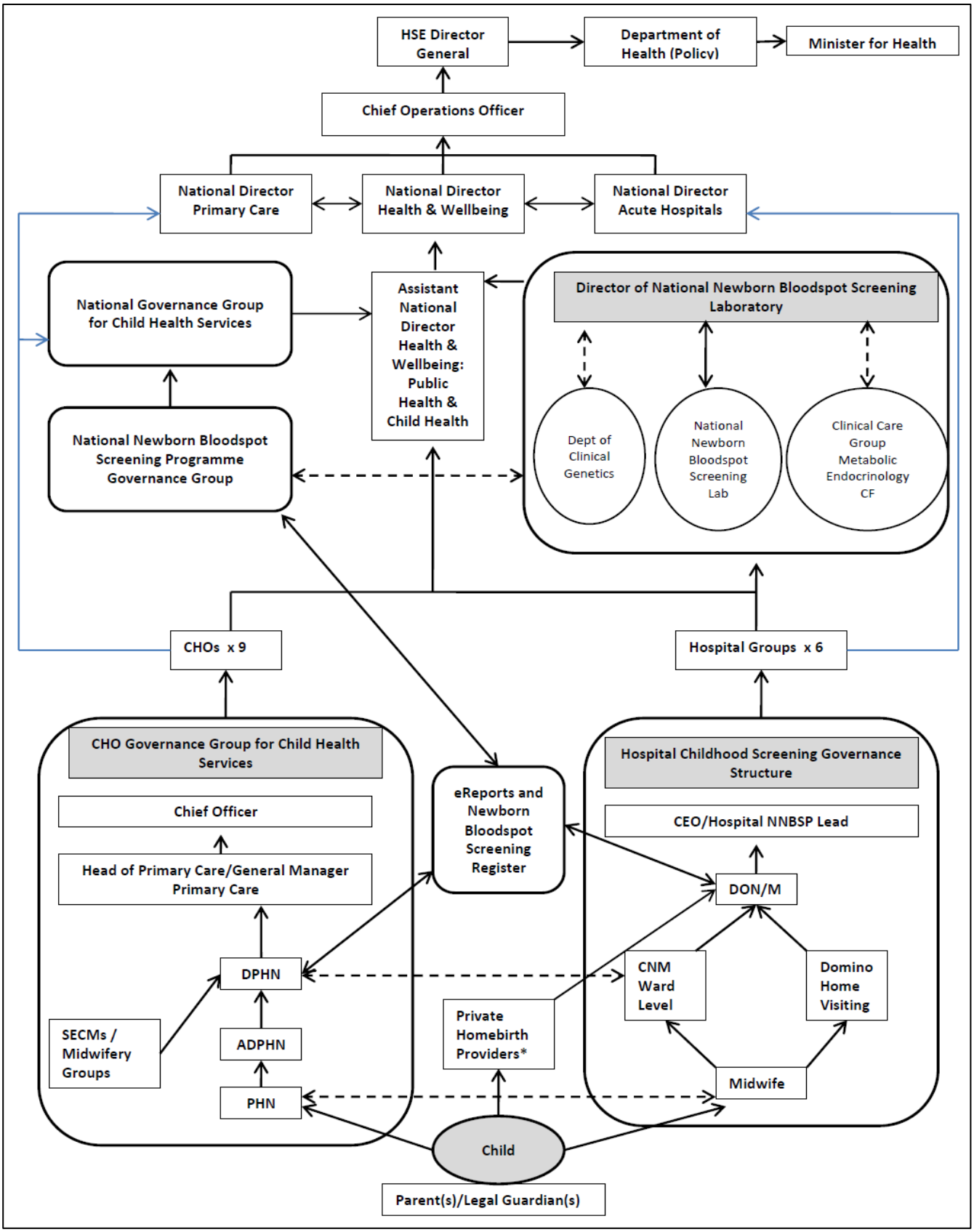
- Locate the baby and parents;
- Explain to the Parent(s)/legal Guardians what disorder the baby is suspected of having;
- Arrange for the baby to attend the designated hospital to be examined, have blood tests performed and a technetium thyroid scan, following which the baby will usually be started on thyroid hormone replacement.

If the blood TSH level is confirmed as being very high and if the baby is approaching ten days of age thyroid hormone replacement may be started before they attend the hospital for a thyroid scan.

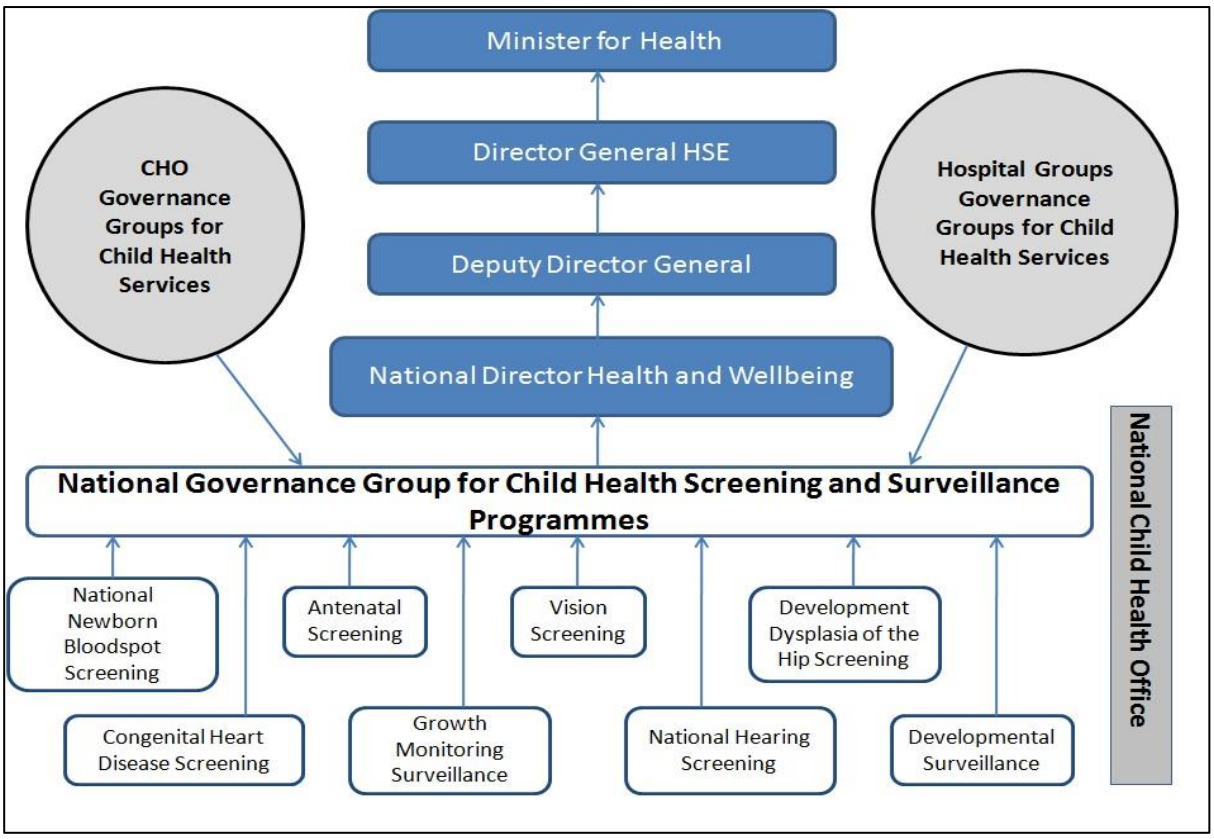
Appendix XXVI: NNBSP Governance Group

Name	Position
Dr. Phil Jennings	Director of Public Health and National Lead National Healthy Childhood Programme (Chair)
Paul Marsden	Project Manager Child Health Screening and Surveillance Programmes
Dr. Abigail Collins	Consultant in Public Health Medicine
Dr. Ingrid Borovickova	Clinical Director, National Newborn Bloodspot Screening Laboratory, Children's University Hospital Temple Street
Ms. Loretta O'Grady	Chief Medical Scientist, National Newborn Bloodspot Screening Laboratory, Children's University Hospital Temple Street
Dr. Ahmed Monavari	Clinical Director, National Centre for Inherited Metabolic Diseases, Children's University Hospital, Temple Street
Dr. Barry Linnane	Paediatric Respiratory Consultant, University Hospital Limerick
Mary Finn Gilbride	Director of Public Health Nursing Wexford LHO
Colette McSweeney	Assistant Director of Public Health Nursing, Cork South Lee LHO
Rita Lawlor	Professional Development Co Ordinator for Practice Nurses/Interim Immunisation Co-Ordinator, Primary Care Unit, Dublin Mid Leinster,
Grace O'Neill	Regional Child Health Training/ Development Officer/Immunisation Co -Ordinator, HSE South East
Barbara Bolger	National Specialist Primary Care Operations, Primary Care Division (Formerly Regional Specialist Primary Care HSE Dublin North East)
Ms. Angela Dunne	Director of Nursing, Midland Regional Hospital Portlaoise and Lead Midwife, National Women and Infants Health Programme
Ms. Jacinta Egan	Assistant Staff Officer, National Healthy Childhood Programme

Appendix XXVII: NNBSP Governance Structure



Appendix XXVIII: National Governance Structure for Child Health Screening and Surveillance Programmes



Appendix XXIX: CHO Governance Structure for Child Health Screening and Surveillance Programmes